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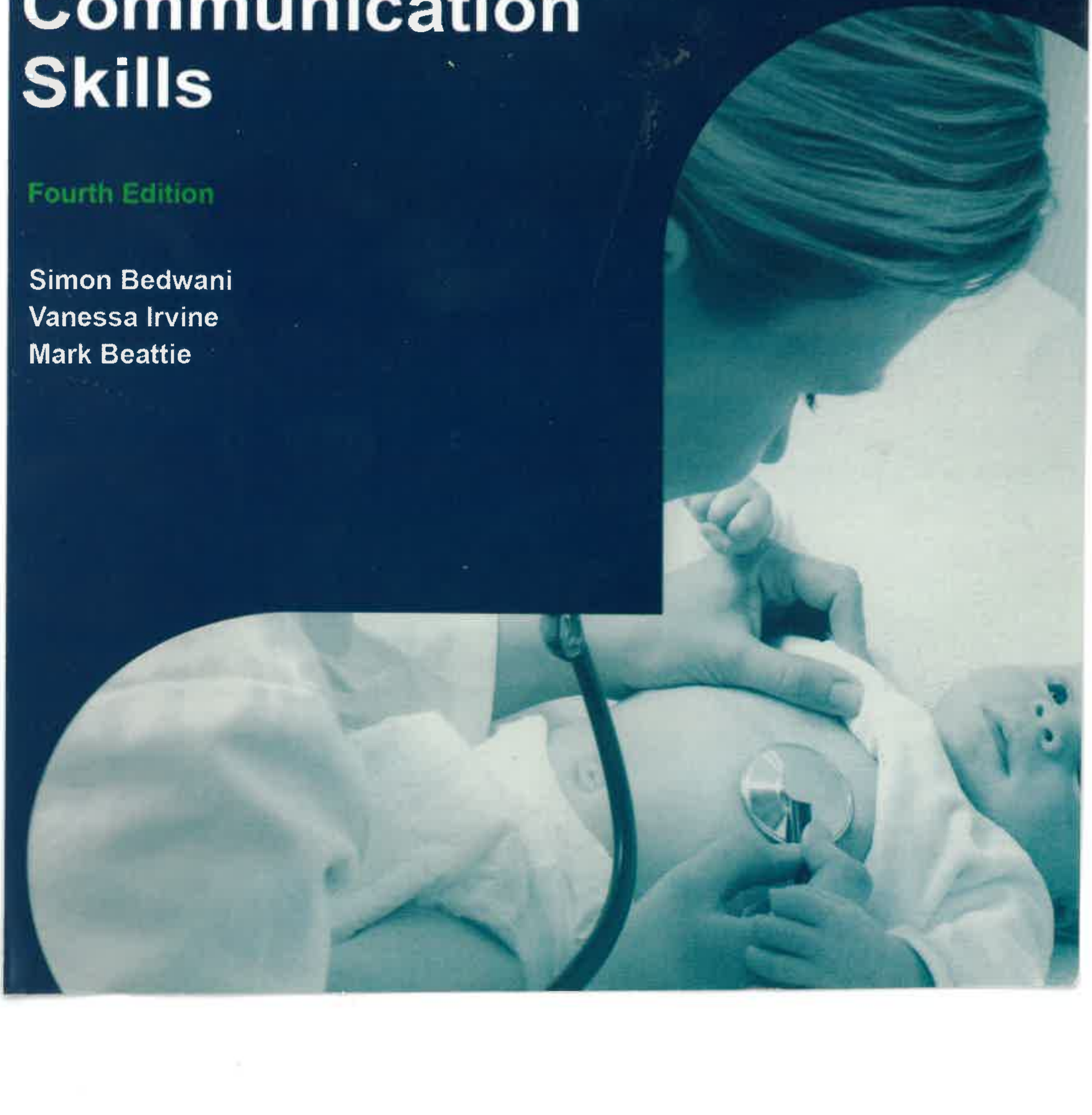
MRCPCH Clinical Short Cases, History Taking and Communication Skills

Fourth Edition

Simon Bedwani
Vanessa Irvine
Mark Beattie



© 2016 Pastest Ltd
Egerton Court
Parkgate Estate
Knutsford
Cheshire
WA16 8DX
Telephone: 01565 752000

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For further details contact:

Tel: 01565 752000 Fax: 01565 650264
www.pastest.com enquiries@pastest.com

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INTRODUCTION

The structure of paediatric training in the UK has gone through some significant changes in the past, and will continue to change over the next few years. What has not changed is the fact that the College examination is still a significant hurdle to the paediatric trainee.

Like the examination, this book is split into three parts – History Taking and Management Planning, Communication Skills and short case examination for the Clinical Stations.

Part I History Taking and Management Planning has been written by first giving practical tips and pointers on taking a good history and conducting oneself appropriately in this station. There then follow some in-depth examples of cases with the emphasis on how the good candidate might tailor their history to answer questions posed in the form of a referral letter, and also mock 'question and answer' sessions to reflect what happens when one is left with the examiners at the end of the station. There are additional cases that can be worked through by the candidate and used as practice questions.

Part II Communication Skills is designed to help the candidate maximise their chances of doing well in this potentially tricky section of the exam. There are suggestions about how to practise for the station, templates helping you to deal with different categories of tasks and worked examples.

Part III Clinical Stations has been split into sections as seen in the examination – Cardiovascular, Abdominal, Respiratory, Neurological, Musculoskeletal/'Other' (encompassing rheumatology, haematology, dermatology, endocrinology and growth, and nephrology) and the Child Development Station. The text includes notes on examination technique, useful background information and summaries of cases that have been seen in recent exams.

We do not cover the video station anywhere in this book. This station tends to assess examination and management of acutely unwell children who could not easily be brought along to the short case station. The conditions seen reflect those that a paediatrician would encounter on call or on the wards, and your exposure to hospital paediatrics will be a good preparation.

This book has been put together by authors who between them have had experience of teaching on membership courses, writing previous Pastest books and also helping to run previous examinations. Much of the advice in this book comes from direct experience of recruiting appropriate patients, briefing candidates, looking after both patients and candidates during the examination and listening to candidate feedback about cases and scenarios encountered.

The book is not a textbook of paediatrics or indeed a textbook on clinical skills but contains as much useful information as possible with the aim of helping the candidate pass the exam. The principles outlined in this book are designed to be applicable on the wards and in outpatient clinics, where every day there are tests in our ability to take a focused history, examine patients and communicate with those around us.

We have carefully and conscientiously endeavoured to provide information in this book which is considered correct and up to date at the time of writing. Our aim is to summarise what we consider to be current practice in a helpful way. It is a medical professional's duty, however, to check their own practice with a variety of sources, including college or local hospital guidelines, policies and procedures as well as peer-reviewed literature.

We hope you find the book helpful.

Simon J Bedwani, Vanessa Irvine, R Mark Beattie
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We would like to thank the following contributors for their work on previous editions:

Christopher J R Anderson
Consultant Paediatrician, Salisbury District Hospital, Salisbury

Andrew T Clark
Consultant in Paediatric Allergy, Addenbrooke's NHS Trust, Cambridge

Anne Smith
Consultant Paediatrician, Northampton General Hospital, Northampton

PREPARING FOR THE EXAMINATION

When it comes to preparing for exams what works for one person won't necessarily work for another. This exam is slightly different; you need to have the background knowledge which will have got you through your three theory exams. However, to pass this exam you need practical experience, lots of exposure to patients with clinical signs and ... lots of practice!

Here is an approach to how and when to start preparing for your exam.

Firstly decide which sitting you are going for and commit to it.

6 months to go

Attending as many clinics (both general and sub-specialist) as possible will really pay off, as will making a point of going to examine inpatients in your department with clinical signs.

Arrange to sit in on a variety of clinics. Realistically when you are preparing for an exam you revise on your days off. It may be that on your days off you 'book in' to go to clinic. It is exactly these types of patients that will end up in your exam!

Start regularly practising doing 'full systems examinations'. This needs to become absolutely fluent and second nature. In your clinical exam you need to be concentrating on eliciting the clinical signs, and not on trying to remember the sequence of the examination.

Start finding out who in your hospital/deanery is sitting the exam at the same time. Find out from colleagues who have recently sat the exam which consultants/registrars would be good to ask for teaching sessions.

Consider booking yourself onto a good clinical course that has been recommended to you.

3 months before the exam

Get your study group together.

Write a timetable and allocate sessions to organise among yourselves. Often the best structure for these sessions is to go and see a patient as a group with the senior doctor leading the session. One person should examine the relevant system. Another should critique, followed by the senior doctor. You can then discuss differential diagnoses/treatment options, etc.

Do not waste these sessions if the consultant/registrar is late/fails to turn up – use this time to observe and critique each other's examination skills and practise presenting your findings. It is really obvious which candidates have practised their presentation of clinical findings; it is not something we do often in our day-to-day practice, and so takes time to get back into the habit of doing this slickly and confidently.

Though role play makes most of us cringe – get over it and go for it! This is exactly what you will be doing in your history taking and communication skills stations. Stumbling awkwardly through a role play or listening to other people role playing can be helpful, and an amazing motivator for you to think how you could phrase things differently!

1 month to go

Keep going!

Continue your group sessions.

This would be a good time to go on a course.

Only 4 weeks to go – stay focused and remember … keep practising.

On the day of the exam

The format and length of the exam mean it can be physically as well as mentally taxing:

- Try and have a day off from revision before the exam and an early night the night before.
- Eat a decent meal before heading to the exam.
- Dress in smart, comfortable clothes that are not restrictive, so you can perform the examinations required (including a developmental assessment which may require getting down onto the floor to get involved!). Remember bare below the elbows.

Plan how you get to your exam leaving lots of extra time, so as not to turn up stressed/distracted.

Once you have arrived it is vital to concentrate on the route that is particular to your centre, and be mindful of the location of toilets, water, etc. Although there will be people there to direct you to the next station, it helps you to concentrate if you can smoothly make your own way there.

When the examination has started, there are some valuable points to remember.

The first is to drink some water at strategic points on your way around the circuit.

The second (based on observations of candidates) is that they have been asked to produce their mark sheet for the examiner at the start of the station but have left it outside the room on a chair. This means they have had to exit the room, pick it up and re-enter. This is mildly embarrassing but moreover breaks concentration and feels like a 'bad start' when you are nervous ...

Throughout the exam, remember to be yourself – being kind and friendly to the examiners/patients/parents will only help!

We hope that if you have prepared thoroughly and followed the advice given to you by your colleagues, patients, parents and by ourselves, you will have ironed out any uncertainties or bad habits.

You will then pass!

GOOD LUCK!

THE MRCPCH CLINICAL EXAMINATION

The Clinical Examination is designed to reflect the skills required to partake in 'middle-grade' rotas and training. The well-established OSCE-style format means that the candidate is observed throughout the examination by a total of at least nine different examiners, with mark sheets based on definable, reproducible criteria.

The aim of the examination is to assess whether the candidate has the necessary **all-round skills** that would be expected of a new specialist registrar.

The RCPCH website (www.rcpch.ac.uk) contains excellent information and links for candidates, including examples of questions and mark sheets. What follows below is a synopsis of the examination with some tips on how to cope on the day.

A clearly defined **circuit** is set up which includes the following stations:

History taking and management planning (22 min)
A test of your ability to take a focused history and discuss a management plan.

Communication skills (two stations; 9 min each)
Primarily a test of good communication but background knowledge does help.

Clinical video station (22 min)
This tests recognition of acute symptoms and signs, many of which cannot be tested in the short cases. Conditions such as severe croup and meningococcal disease are examples. The video station will not be mentioned again as it is

beyond the scope of this book to provide more detail about this part of the examination.

Clinical stations (six stations; 9 min each)
A series of 'short cases' testing physical examination skills, interpretation of findings and knowledge around the case in general.

You can start at any point on the circuit. There are 4-min breaks between each station with the entire examination lasting 152 min. Candidates can see the circuit diagram on the RCPCH website and should be shown around their examination circuit on arrival at the exam centre.

PART I

History taking and management planning station

1

INTRODUCTION AND SUGGESTED APPROACH

The aim of this station is to assess your ability to take a focused history, be able to formulate a management plan, and then summarise back to the examiner the main issues pertinent to the case.

This station closely reflects an outpatient or ward review setting, in keeping with the philosophy of the exam. Children could have a new diagnosis or presenting complaint, or you may be asked to address a specific problem in a child with a known diagnosis. You will never be asked to examine a patient but may be given additional information, eg results of investigations.

You will be given instructions that provide information about your role. These are often in the form of a referral letter. The parents and child will also have been briefed; in many cases the clinical scenario or at least the child's background problem is real. In some cases a role player, health care professional or member of the public may be used.

This 22-minute station is divided into 13 minutes with the patient (with a 2-minute warning) and 9 minutes alone with the examiner to discuss the case.

The 2-minute preparation time before the station is of crucial importance as it gives you time to gather your thoughts about the case. After all, nobody would want to ask a new patient into a clinic consultation without having read the referral letter thoroughly!

The Royal College of Paediatrics and Child Health (RCPCH) Anchor Statements for Expected Standard/Clear Pass demonstrate that three key areas are assessed:

- Conduct of interview (ie your behaviours and attitude),
- History taking,
- Interpretation/management planning.

These can be found in the Examinations section of the RCPCH website (www.rcpch.ac.uk/Examinations/MRCPCH-Clinical), and are summarised below.

1.1 Conduct of interview

- Greet parents and child appropriately and introduce yourself
- Clarify your role and agree aims and objectives
- Maintain appropriate eye contact/body language throughout
- Remain attentive throughout; give verbal and non-verbal signs to that effect
- Make sure you are fully understood by patients, parents and examiner
- Be confident yet empathetic
- Patient and parent(s) should be at ease with you

You may feel that all of the above behaviours apply to you in a work setting but how many times have you been observed? What feedback have you received from consultants, peers, nursing staff, parents and children? A little reflection will not go amiss when planning for this section of the examination. The day of the exam is not the best time to discover that you actually talk with your hand over your mouth or look at the floor when you are anxious! The greeting is all-important, as are your clarity of speech, eye contact and general approach throughout the station.

1.2 History taking

Doing this well involves:

- Phrasing questions clearly and making sure they are not all 'closed'
- Avoiding more complex medical terminology that may not be understood by the patient/parent
- **Allowing child/parent time to speak** – this takes conscious effort in the stress of an exam setting
- Developing a problem list as you go
- Exploring the main problems fully, including psychological, social and family issues as these may be what you are asked about
- Ensuring your history is focused to getting maximum information
- Exploring feelings of the patient and parents – ideas, concerns and expectations
- Concisely summarising the above information in a structured way

ALL THIS NEEDS PRACTICE

Practising your history taking skills will reap dividends in the exam and will reveal any faults that may have become ingrained over the years. It is particularly important to let parents/patients speak, as this will reveal many issues without your having to ask. Many of us don't pause for the answer to the question, particularly when we are feeling nervous or rushed. Get into groups and test yourselves on this specific issue.

The skilled inteviewer knows when to use open and closed questions and to interject politely when sufficient information has been required in order to move on and explore another area. The different headings and questions used in the examples that follow are a guide to the information to be elicited – you may obtain the answers quite organically without having to ask a series of closed questions!

Just like the Clinical Section, you will need to get used to being scrutinised while you perform the task in hand. You will have undertaken a few Mini Clinical Examinations (CEXs) so you should already be used to being watched while taking a history. What were your strengths and weaknesses in those situations? If you haven't done this too often before, ask your colleagues (of any grade) to watch your patient reviews and comment on your 'performance'.

1.3 Interpretation and management planning

○ Summarise the case to the examiner – it is very important to be able to summarise a case succinctly without going through every detail again; this will get your 9 min with the examiner off to a good start and boost confidence. Look at your previous entries in clinical notes to see how often you write a brief impression or summary of a case as well as a sensible differential diagnosis – getting used to doing this will help in the exam and improve your performance during handover and on ward rounds

○ Demonstrate a good understanding of different problems raised in the history

○ Discuss the differential diagnosis listing the most likely first – not some rare differential, especially if you cannot talk about the condition you have just blurted out while under pressure!

○ Outline the essential points in your proposed management plan; be sensible and pragmatic. Do not suggest any experimental or controversial treatments unless asked about them directly. There are many national guidelines on the management of common conditions that are referenced in this book and easily available through a simple web search. Your local Trust/network guidelines would also be a good place to look.

○ Ensure that your management is appropriate and effective, and addresses patient/parent concerns that have been raised in the history

- Discuss referral to other agencies and be aware of providing both written information and further contact information, eg parent groups/respite – this is often forgotten or done poorly. If you cannot remember organisations for specific illnesses, Contact a Family (www.cafamily.org.uk) is a good place to start for information on many chronic conditions.

A thorough knowledge of not only the medical but also the psychosocial aspects of many chronic conditions is essential. There are many common principles such as the need for multidisciplinary support, the need for consideration of education/family issues and other psychosocial factors.

Try to list these kinds of issues for each condition – they often have common themes and may not match the problems you will read about in many standard texts.

SUGGESTED APPROACH

The following is a suggested template that you can apply to most cases and will be used in the six full examples that follow in Chapter 2. You still need to be fluid in your approach as things may take an unexpected turn.

1.4 Preparation time – 2 minutes

Read the candidate information and the task itself (often a referral letter) thoroughly, including the background information. There may be clues as to which avenue to take in the consultation. Imagine yourself in this role and setting. Try to think of an opening line after you have greeted the parent and child. What are the main issues you wish to clarify about this case? What particular concerns might the parent have, eg potential side-effects of a new treatment?

Try to put yourself in their position for a moment; what would be worrying you most as a parent? You are allowed to make notes and bring them in with you.

1.5 Consultation – 13 min

First 9 min

Introduce yourself, explain your role. Ensure that the patient and parent are comfortable and at ease.

Outline the aims and objectives of the consultation – best done by initially referring to the letter.

Proceed to take a focused history.

You can use the template below but home in on the most important problems. This may mean that you cannot cover all of the headings below – try not to feel too uncomfortable about this.

Remember, the RCPCH guidance notes stipulate that this 'is not a test of the ability to take a comprehensive history.'

- Presenting complaint:
    - History of presenting problem(s) – including descriptions of symptoms and what has been done so far. If the child has a chronic condition this is a good place to explore the background history of that particular condition, eg how it was diagnosed and when, GP visits/hospital admissions/professionals involved so far
- Pregnancy (where relevant), birth history, neonatal history
- Past medical history including relevant previous illnesses – could ask, 'Any previous illnesses, admissions, operations?'
- Development
- Immunisations
- Drug history – remember to include doses and compliance
- Allergies – if reported drug allergy try to get concise description of what happened
- Social and family history – try to explore whether there are particular concerns/difficulties. Examples may be one-parent family/parental illness/transport difficulties/partner away most of time/siblings with same problem, etc.

Explore support systems that the family might have in place. Are you actually talking to the parent who knows the child's illness best?

- Brief review of systems to ensure you haven't missed out something important

Last 4 min

The bell will ring signifying that there are 4 minutes remaining with the parents and child. At this stage you should be thinking of exploring any concerns, expectations and feelings that have not come out of this consultation so far. Do you feel there are issues that have not been covered?

It is useful to ask such questions as:

- What is your biggest worry?
- Are you concerned about any diagnosis in particular?
- Is there anything else you would like to tell me/ask me?
- Is there anything you haven't understood?

When you feel that the consultation has come to a close you should summarise the main points back to the parent and child as appropriate. This might result in an additional useful nugget of information.

1.6 Reflection – while parent and child are shown out of the room

Do not panic; try to develop a problem-orientated summary of the key facts/issues of the case. Do not reflect on the information that you could have gathered but didn't – be positive.

1.7 The 9 min with the examiner

Summarise the case, including the main issues covered in the history. Come up with responses to potential issues, a differential diagnosis, investigations and management plan. Suggest contacts for the family and outline how you would refer the patient to other agencies, where relevant.

2

CASES

CONTENTS

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2.5 Case five: Cerebral palsy
    Notes on management of cerebral palsy
2.6 Case six: Nephrotic syndrome – relapse
    Notes on nephrotic syndrome

In this chapter there are six full example scenarios. For each of the scenarios you are given a **role**, a **setting** and a **task**. There will also be some **background information**.

Remember that the examiner will have been given information on some key features in the medical and social history that you will be expected to elicit.

The first case includes initial instructions that would be similar to the candidate instructions seen on the examination sheet. See also the example question on the RCPCH examinations website, within the PDF file 'MRCPCH Clinical – information for candidates'. It is good practice to familiarise yourself with the timekeeping instructions in the Box. These are not given for the other five full examples to avoid unnecessary repetition.

A template for how you might approach the history is also given, along with some information that a good candidate would elicit from the history and that the examiner will have to hand. Note that some of the information is not specific to that condition but is very relevant – especially the social situations covered.

Some notes on management pertaining to the scenario in particular (using a question and answer format) are then provided. These are followed by additional notes on the condition/presentation, as there are of course many possible scenarios for a given condition.

A further 12 examples of scenarios you may encounter are given in Chapter 3, along with more concise guidance about the most important information to elicit, and key management points. These can also be practised as full scenarios with colleagues.

2.1 Case one: cystic fibrosis – respiratory deterioration

- Take a focused history having read the scenario below
- The subject/role player may ask you questions
- When the consultation has finished the examiner will discuss your management plan with you

Timing:

*Total time provided for this station is **22 min**.*

*There are **2 min** before you are called in to read this sheet and prepare yourself. A bell will signal the start of the station and you will be invited into the examination room – take your instruction sheet with you.*

*You will have **13 min** in total with the patient; a warning will be issued when you have **4 min** left to complete your history*

*You will then have **9 min** alone with the examiner to discuss your management plan.*

You may take notes.

You will not be asked to examine the patient.

Role

You are the Specialist Trainee.

Setting

Cubicle on the children's ward of a district general hospital.

You are talking to

Craig, a 10-year-old boy, and his mother.

Task

Take a focused history, aiming to explore the problem indicated as you would in the clinical situation. You may answer questions that the subject (role player) may pose to you. After the consultation the examiner will focus on your management planning.

Dear Dr...

Re: Craig S

This boy, who is 10 years old, has cystic fibrosis. His family are visiting the area during the half-term holiday, and the boy was last seen in his usual clinic 6 weeks ago for his annual review. He is colonised with *Pseudomonas*. He has become very chesty over the last few days and is a lot more tired. On examination he has a temperature of 37.8°C, and on chest auscultation he has coarse expiratory crackles at the right base. I would be grateful if you could assess him; I do wonder whether he needs admission.

Yours sincerely,

Dr S Smith

General Practitioner

Background information

Craig has been seen regularly in his usual cystic fibrosis clinic at City Hospital (his CF Centre) and has had very few admissions as an emergency. He has not attended any other hospital before now.

Any other information

Your examination findings are the same as Dr Smith's.
Craig is thin (his weight is on the 0.4th centile).

Preparation time (2 min): initial thoughts

Once you have read the question, concentrate on remembering the child's name and coming up with an appropriate greeting to him and his mother. What issues can you see between the lines already? In this case the family are not used to this particular hospital and might feel less confident than they normally would.

Consultation

Introduction. Clarify role, agree aims and objectives.

Remember to include the patient as much as possible in the consultation, which is sometimes difficult when the parents talk freely and you are nervous.

History

- When did Craig become chesty?
- What are the main symptoms? Ask about fevers, malaise, wheeze
- Triggers if wheezy episodes
- Exercise tolerance at moment – compare to normal
- Results of last lung function tests
- Results of last sputum sample or cough swab – GP says *Pseudomonas*.
- Number of hospital admissions per year – elective and emergency
- How was Craig diagnosed?
- Nutritional issues/gut symptoms:
  Has Craig been gaining weight along 0.4th centile or is he losing weight? Ask about stool frequency. Has the dose of pancreatic enzyme supplementation needed adjusting recently?
- Other health professionals involved, eg physiotherapist, dietician
- Birth and neonatal history – *brief enquiry in this case*
- Any other medical problems?
- Immunisations, especially for *Pneumococcus*
- Drug history. Any problems with compliance?
- Allergies – particularly to antibiotics
- Family and social history

Warning bell rings; you have 4 min left with Craig and his mother. You have gathered the following information so far:

Craig has had a cough for the last 5 days, which has gradually worsened in frequency. He has become productive of green sputum and developed a fever today. He can still run around but only for 10 or 15 minutes; he then needs to sit down.

He was diagnosed with cystic fibrosis as a neonate having had meconium ileus which necessitated surgery on day 10 of life.

There have been many elective admissions for antibiotics over the years, which have needed to be more frequent. He had an implantable venous access system (Port-A-Cath®) inserted 6 months ago. On his last clinic visit lung function had deteriorated and sputum was colonised with *Pseudomonas*. He has always been close to the 0.4th centile for weight. He is not nasogastrically fed overnight.

Medications: oral ciprofloxacin, multivitamins, Creon® pancreatic enzyme supplements.

Craig lives with his parents and 4-year-old brother, who is fit and well. This is their first holiday with Craig for 3 years as they have been worried about being too far from their local hospital.

Questions to ask at this point:

○ Who is Craig's consultant?
○ Who is looking after Craig's brother and where are you all staying at the moment?
○ Is there anything that you would like to ask me? Remember to address this to both Craig and his mother
○ Have you any other concerns that we have not covered?

It transpires that Craig's mother was extremely anxious about coming away with him, as he has not had such a good year. They are staying with his aunt who is currently looking after Craig's brother. The father is back home at work. To compound matters, they forgot his PEP mask (Positive Expiratory Pressure device used as part of the daily physiotherapy routine to aid clearance of secretions) while packing for their holiday and she feels this is the cause of this deterioration.

You verify the main points and summarise the history. There are no further questions from Craig or his mother.

The bell rings and you are left with the examiner.

Discussion with examiner (9 min)

Would you please summarise this case?

I saw Craig, who is 10 years old and has an infective respiratory exacerbation of cystic fibrosis. He is visiting the area, and is normally looked after by the CF team (Dr Jones) in City Hospital. He is colonised with *Pseudomonas,* and his background respiratory function has been deteriorating recently. He has also developed an episodic wheeze. The family are additionally anxious as they are away from home (and their CF centre) for the first time since his diagnosis.

How would you manage this child's current exacerbation?

Points to cover here are:

○ Hospital admission (cubicle), saturation monitoring, oxygen as needed
○ Investigations: cough swab or sputum sample, blood cultures and inflammatory markers, peak flow, lung function tests if available
○ Find out results of last cough swab/sputum by contacting his usual centre. When was his last chest X-ray? Have tests for *Aspergillosis* been performed recently?
○ Treatment with iv antibiotics such as ceftazidime and tobramycin (may be worth discussing with your local microbiologist)
○ DNase

- Physiotherapy twice a day; liaise with department and give them a summary of Craig's history.

Craig's mother wants to go back to the City Hospital as soon as possible. She wants to see the cystic fibrosis team, and her other child goes back to school in 2 days. What do you tell her?

... that you will liaise with your consultant and the local cystic fibrosis team to try to transfer Craig back to City Hospital providing he is well enough for the transfer as soon as possible. He will require a cubicle. He will be here overnight at least and we will discuss our management plan with his cystic fibrosis team.

Notes on some important clinical problems in cystic fibrosis

Respiratory

- Almost all deaths in cystic fibrosis patients are due to pulmonary disease. As well as bacterial colonisation and infection, cor pulmonale and allergic bronchopulmonary aspergillosis (ABPA) need to be considered
- Cor pulmonale can develop in the second decade secondary to severe lung disease; it is recognised clinically by a raised jugular venous pressure, hepatomegaly and oedema. Confirmation is on echocardiogram. Mean survival subsequently is 8 months
- Allergic bronchopulmonary aspergillosis (ABPA) occurs in up to 10% of patients: wheeze, eosinophilia, raised specific IgE and IgG and/or positive sputum culture for *Aspergillus*
- Asthma: up to 20% of patients have atopic asthma

Gastrointestinal/hepatic

- Pancreatic insufficiency with resultant malabsorption in 85% of patients – resultant failure to thrive, steatorrhoea
- Meconium ileus in the neonatal period is the presenting feature in 10%–15% of patients
- Distal intestinal obstruction syndrome (DIOS) or meconium ileus equivalent occurs in older children (20%), presenting as cramping abdominal pains and a palpable mass in the right iliac fossa. Remember that such masses may form the apex of an intussusception
- Rectal prolapse occurs in up to 20% (toddler presentation) and is caused by a combination of steatorrhoea and coughing. It usually resolves on starting pancreatic replacement therapy
- Patients may present in the neonatal period with obstructive jaundice due to inspissated bile

- Chronic liver disease occurs mainly in teenagers (around 20%); some develop focal biliary cirrhosis and 2% of these progress to multilobular cirrhosis with portal hypertension and oesophageal varices. For this reason there should be annual screening for emergent liver disease in the over 5s (checking LFTs, clotting, USS)

Endocrine

- Insulin-dependent diabetes, also termed CF-related diabetes, occurs in 7% of paediatric patients (mainly over 10 years), secondary to pancreatic fibrosis. It should be considered in those who have CF and develop unexpected faltering growth or weight loss. It can be easily controlled with the timely introduction of insulin, and diabetic ketoacidosis is rare. However, as patients survive longer, the incidence increases, and evidence of microangiopathy and subsequent complications is now emerging.

Metabolic

- Salt deficiency state, especially in summer months (loss through sweat), resulting in hyponatraemic, hypochloraemic metabolic alkalosis. Potentially major problem in active young patients who still rely on parents for drinks
- Reduced bone mineral density can occur – should be screened for using DEXA scans in the over 10s

Genitourinary

- Azoospermia and so male infertility

Psychological

- Particular problems such as needle phobia, disordered eating
- Acceptance of a chronic, life-limiting condition and compliance issues
- Difficulties with transition
- Depression and anxiety should be considered.

Notes on management of cystic fibrosis

Emphasis is on multidisciplinary outpatient-based management, involving medical staff, physiotherapist, dietician, community nurses, social workers and psychologist as required. Annual specialist review should be undertaken. Please also refer to the Standards of Care consensus documents on the Cystic Fibrosis Trust website (www.cftrust.org.uk), a useful source of information. Many aspects of care are covered, including antibiotic treatment, physiotherapy, infection control and nutrition.

Physiotherapy

There are several airway clearance techniques, often used in combination. Examples are the active cycle of breathing technique, postural drainage, use of a positive expiratory pressure (PEP) mask/oscillatory device (eg flutter device), to name but a few.

Airway clearance can be aided by inhaled hypertonic saline nebulisers.

A regular exercise programme is also beneficial, and has been shown to improve lung function alongside physiotherapy.

Nutritional management

Patients with cystic fibrosis suffer because they have increased metabolic demands, malabsorption and poor appetites. Assessing and optimising nutritional status is one of the most important factors in managing a patient with cystic fibrosis as this is strongly correlated with the severity of pulmonary disease. All patients are referred to a dietician and should be managed on a high-calorie, high-protein diet. This can be supplemented by high-calorie drinks. Overnight nasogastric feeds are an alternative as appetite is often poor, and eventually some patients may benefit from overnight gastrostomy feeding. Vitamin A, D and E supplementation is necessary as these are fat soluble. Plasma concentrations should be measured at the annual review.

Patients should have their height and weight and BMI plotted on centile charts every 3 months. Infants should also have head circumference measured and plotted.

It is important not to provide excessive nutrition, and avoidance of obesity, particularly with additional enteral feeds, needs to be considered.

Pancreatic enzyme supplementation

Most patients will need pancreatic enzyme supplements (pancreatin) in pH-sensitive polymer capsules. The dose is titrated against growth and steatorrhoea. Capsules with a high lipase concentration have been shown to cause colonic strictures.

Antibiotics

Used to reduce colonisation *and* to treat acute chest infections. Colonisation is usually with *Staphylococcus* and *Haemophilus* in infants and *Pseudomonas* in older children. Flucloxacillin is used either prophylactically from diagnosis or long term after colonisation. Nebulised tobramycin/colomycin can be used in *Pseudomonas* colonisation; this has been shown to improve lung function.

Acute infections are treated as they occur. Organisms can normally be predicted from previous cough swabs/sputum samples or isolated. Oral (eg ciprofloxacin) or intravenous (eg ceftazidime/piperacillin plus an aminoglycoside such as tobramycin) therapy can be started as appropriate. Patients have altered pharmacokinetics and so higher doses are used for longer duration. *Burkholderia cepacia* is a highly resistant Gram-negative organism that can spread from patient to patient. Some patients with *Burkholderia* infection can develop a life-threatening septicaemia. Meropenem is often used for this infection.

DNase

Recombinant human DNase. Acts to digest viscous DNA derived from inflammatory cells in bronchial exudates. Most can expect some improvement in forced expiratory volume in 1 second (FEV1) in the first week. Warn patients that it can make them hoarse.

Treatment of co-existent conditions/complications

- Asthma – at least 20% of cases have atopic asthma and should have a written management plan for maintenance and emergency treatment
- Allergic bronchopulmonary aspergillosis (ABPA) – high-dose corticosteroids
- Arthritis – non-steroidal anti-inflammatory drugs (NSAIDS), prednisolone
- Diabetes – insulin
- Constipation/DIOS – laxatives and hydration if mild, through to Gastrografin® or even surgical treatment
- Liver disease – ursodeoxycholic acid used in patients with raised transaminases or early ultrasound features of liver disease
- Cor pulmonale – heart–lung transplant (adults with cystic fibrosis have 70% survival at 1 year, 40% at 5 years).

Separation of patients in clinic settings

This is done to reduce spread of harmful organisms such as *Burkholderia cepacia*.

Psychological and social support

Access to a clinical psychologist is very important, and social workers can also play a role in supporting the family. Play therapists can offer help around procedures in particular, which can be a great help for emerging needle aversion.

Remember the importance of transitioning an older teenager to adult services, eg joint clinics with adult CF team.

Gene therapy

Using vectors such as liposomes to transfer normal cystic fibrosis transmembrane conductance regulator (CFTR) into host cells. Still in trial stages; don't mention unless asked.

2.2 Case two: asthma

Role

You are the Specialist Trainee.

Setting

Outpatient clinic.

You are talking to

Adam, a 12-year-old boy with asthma, and his mother.

> Dear Dr…
>
> Re: Adam J
>
> This 12-year-old boy has attended my surgery five times in the last 3 months with moderate exacerbations of his asthma. He has recently moved house and the family thinks this is the reason for his symptoms. His last attendance resulted in me doubling his flixotide to 100 micrograms twice a day. I have not started any other medications. I would be grateful for your help as he is going through many repeat prescriptions of salbutamol.
>
> Yours sincerely
>
> Dr D McCann
>
> General Practitioner

Background information

Takes occasional cetirizine for hayfever.

Any other information

Weight and height on 50th centile.

Preparation time (2 min): initial thoughts

You should already be thinking of focusing your history to elicit the reasons for Adam's recurrent attendances to the GP.

Consultation

Introduction. Clarify role, agree aims and objectives.

Remember to ask *Adam* about his symptoms. Ask an open-ended question at the start – it engages him in the consultation and may bring out useful information.

Children very often reveal additional triggers of their asthma if allowed to talk by their parents and history taker!

History

- ❍ When diagnosed
- ❍ Number of GP and hospital visits in last year. Known to asthma nurse in GP practice?
- ❍ Number of visits for repeat prescriptions of beta-agonist
- ❍ Number of steroid courses in last year
- ❍ Worst attack and level of treatment needed, eg high dependency unit (HDU) and intravenous aminophylline or salbutamol
- ❍ Description of the last few attacks; are they all related to chest infections?
- ❍ Best peak flow – is a diary kept?
- ❍ Triggers – cover parental smoking, pets, rhinitis, house dust mite, recent house move
- ❍ Other atopic features? May have (had) eczema, food allergy, rhinitis
- ❍ Birth history, relevant past medical history, breast or bottle fed – *brief enquiry*
- ❍ Medications (current and previous), means of delivery and compliance issues
- ❍ Family history of asthma/atopy in general
- ❍ Social history – details of housing, type of flooring, furnishing, pets, measures to reduce house dust mite numbers
- ❍ Systems review (can sometimes pick up side-effects of medications, eg tremor from excessive salbutamol use, headaches as anxious about worsening condition/bullied in school)

Warning bell rings; you have 4 min left with Adam and his mother. You have gathered the following information so far:

Adam was diagnosed at 6 years of age following admission to hospital for severe wheeze requiring admission to the HDU and intravenous salbutamol. He has also been seen by a previous GP for viral-induced wheeze and eczema between the ages of 1 and 2 years. In retrospect his mother thinks he had some mild exercise-induced symptoms before this first admission. He was seen in clinic for a couple of years then discharged as he was stable on 100 micrograms of flixotide per day. Adam is becoming more independent and is taking the flixotide without supervision. He has not been given any other medications. He has been worse in the last few weeks, taking his salbutamol two to three times a day. He has required four courses of prednisolone in the past 5 weeks. The family have moved house recently and there is a lot of building work. His father is quite stressed and has taken to smoking again; he had given up for a few years after Adam's first admission.

Questions to ask now:

○ How exactly is he taking his flixotide? Does he use a spacer?
○ Ask Adam about compliance. If you phrase this question correctly you are more likely to get an honest answer. For example, asking 'do you take your flixotide Adam?' will result in 'Yes'. Asking the same patient, 'Adam, do you ever forget to take your flixotide? Lots of people do when they are in a rush' will also result in a 'Yes'! Then gently ask how often he misses it.
○ Explore parental knowledge about inhaled steroids and risk factors for worsening asthma such as parental smoking (if time).
○ Has he settled into his new school?

Adam tells you that he has been missing all his morning doses of flixotide recently because he has started walking to his new school; he has to set off earlier and he finds it hard to remember when he is in a rush. Besides, he doesn't notice any obvious benefits when he takes his flixotide – the salbutamol is better. When he remembers, he takes his flixotide using an Easibreathe®. He enjoys school.

You verify the main points and summarise the history. There are no further questions from Adam or his mother.

The bell rings and you are left with the examiner.

Discussion with examiner (9 min)

Would you please summarise this case?

Adam is a 12-year-old boy with a long history of asthma and past history of eczema, but no history of rhinitis. One of his exacerbations 6 years ago required iv salbutamol on HDU. His control has worsened recently and this coincides with a recent house move, less good compliance and exposure to passive cigarette smoking.

What other reasons might there be for poor response to his current asthma therapy?

The Easibreathe® device may not suit him. He may also need additional medication.

How would you manage this boy now?

Cover the following:

- Educate parent and child about the importance of taking inhaled steroids regularly and their mechanisms of action. Mother will have to supervise for a while longer. You could tell him to take it before he brushes his teeth morning and night
- Check inhaler technique. A large volume spacer would be more appropriate for flixotide, especially while control is not as good (see NICE guidelines on inhaler devices in children 5–15 yrs at http://guidance.nice.org.uk/TA38/Guidance)
- Encourage use of peak flow diary; make sure there is a written management plan for future attacks. Plot his predicted peak flow for height
- Address paternal smoking; ask GP to help in your clinic letter if necessary
- Optimise the management of his hayfever. Remember that children can still have significant rhinitis over the autumn and winter months, particularly if they are house dust mite-allergic.
- Leave other medications as they are and review in a few weeks; they may have an effect if taken regularly. If control is still poor, the flixotide dose can be doubled to 200 micrograms/day. If subsequent control is still poor, and the patient is compliant, add in a long-acting beta-agonist or a leukotriene receptor agonist

Notes on management of asthma

The aims are:

- Control of symptoms (including nocturnal and exercise-induced)
- Prevention of exacerbations
- Achievement of normal physical activity and growth along with best possible lung function … with minimal side-effects.

General measures

Avoiding cigarette smoke is the most significant measure. There is no doubt that exposure to tobacco smoke in the home contributes to the severity of childhood asthma. You may be instructed to counsel a proxy parent about smoking – mention the risk to themselves and their child's health as well as offering appropriate support. This reflects the skills you will need in a ward or clinic situation. Remember that a significant percentage of older children smoke.

Weight reduction is recommended in obese patients (an increasing number) with asthma.

Measures to reduce house dust mite numbers, such as washing bedding at 60 degrees, 'anti-allergy' mattress covers, changing carpeted bedroom floors to laminate flooring, *may* reduce symptoms in some patients but these measures are not formally recommended in the British Thoracic Society (BTS) guidelines (www.brit-thoracic.org.uk).

Avoiding animal dander can be difficult for patients with current or even previous pets – even if dogs/cats are removed from a household, their dander will remain in significant quantities for at least 6 months or more.

People often forget to ask children and teenagers where else they go apart from their own home; grandmother's smoky flat and three cats may not feature in your social history but could be the reason for a deterioration.

It is important to explore issues relating to compliance, and to explore psychosocial factors – new school, family bereavement, house move, etc.

Drug therapy

Keep up to date with joint BTS and Scottish Intercollegiate Guidelines Network (SIGN) guidelines. These can be found at www.sign.ac.uk/guidelines/index.html and at www.brit-thoracic.org.uk. Be aware of any changes by the time you take the exam; the latest *British National Formulary for Children* is also a useful reference.

Acute attacks: inhaled or nebulised beta-2 agonists, and oral steroids. Nebulised magnesium sulphate can also be used.

More severe acute attacks are managed with intravenous **salbutamol/aminophylline, hydrocortisone** and in some instances, intravenous **magnesium sulphate**. Ventilation is occasionally required.

Long-term treatment – summary of BTS stepwise therapy as an aide-mémoire:

○ **Step 1**: inhaled short-acting beta-agonist as required
○ **Step 2**: inhaled prophylactic steroids:
  - Consider starting when symptomatic or *correctly and appropriately* using inhaled beta-2 agonists three times a week or more
  - Start at dose appropriate to the severity of disease and titrate to lowest dose at which effective control of asthma is maintained (tailor this dose to the specific steroid using BNFc)
  - The *device* selected for drug delivery is important and age specific. It is important to be familiar with all the inhaler and spacer devices and the most appropriate age to use them (see 'Summary of delivery devices' Box)

  - Remember that the side-effects of inhaled steroids are a common concern to parents and clinicians
- **Step 3:** add-on therapy should be considered if control is not achieved on escalating doses of inhaled steroid, having checked compliance, inhaler technique and triggers:
  - *Long-acting beta-2 agonists* (LABA), eg **salmeterol**, are recommended in the *over 5s*, especially if there are exercise-induced symptoms. Note that they should not be used without inhaled corticosteroids; combined preparations are common (eg **Seretide**).
  - *Leukotriene receptor antagonists* (eg montelukast) may also be added in the over 5s and are used at Step 3 instead of LABA in 2- to 5-year-olds
- **Step 4:** same as Step 3, plus an increase in the dose of inhaled steroid up to 800 micrograms/day in children over 5 years
- **Step 5:** as Step 4 plus oral corticosteroid, plus refer to respiratory paediatrician if have not already done so.

Note that for severe cases, monoclonal anti-IgE treatment (Omalizumab) may be considered. Can increase the number of symptom-free days for children who have severe, persistent asthma with a strongly allergic component. Given by subcutaneous injection in a hospital setting for eligible patients (defined by NICE) every 2–4 weeks. A major potential risk is anaphylaxis.

Summary of delivery devices

Remember you may be asked to demonstrate their use.

- Metered dose inhalers
- Spacer devices: Volumatic®/Aerochamber plus® (+ face mask for < 3s)
- Dry powder: Diskhaler®, Turbohaler®, Accuhaler®
- Self-actuating with propellant: Autohaler®/Easibreathe®

Reasons for poor response to escalating asthma therapy

Remember these when assessing any child with asthma:

- Poor inhaler technique or inappropriate device for that child
- Poor compliance to treatment regimen
- Poor understanding of disease management
- Co-morbidities including emerging allergies (may be new environmental triggers, eg moved house, new pet, building work at school)
- Stress; physical or psychological
- Dysfunctional breathing (eg vocal cord dysfunction)
- Severe 'resistant' form of the disease
- Wrong diagnosis – revisit the diagnosis using BTS definitions.

Summary of assessment of an asthmatic child ready for discharge

This may also be tested in the Clinical Station or even the Communication Station.

Remember to include the following points:

- ○ Adjust the child's medication if appropriate – did under-treatment precipitate the admission, is the child's bronchodilator usage high suggesting poor control (contact GP – how many repeat prescriptions?) and is there a need to increase prophylactic medication?
- ○ Check patient is stable on the medication they will receive at home; should not need bronchodilator more frequently than 4-hourly
- ○ Check inhaler/spacer and peak flow technique and adjust as necessary
- ○ Explain in basic terms the difference between bronchospasm and inflammation and the relevance of using a regular preventative steroid in that context
- ○ Provide a written action plan with clear instructions of what to do if the asthma worsens, and provide a peak flow diary
- ○ Give advice on avoiding precipitants
- ○ Write to the GP (timely discharge summary) and ask them to review the child soon
- ○ Arrange community or asthma nurse to visit, eg if there are concerns over compliance
- ○ Arrange follow-up in clinic if appropriate.

2.3 Case three: ex-premature baby with recurrent wheeze

Role

You are the Specialist Trainee in a district general hospital.

Setting

Outpatient clinic.

You are talking to

Jess, mother of Samuel.

Dear Dr...

Re: Samuel Adams

Thank you for seeing this 14-month-old boy with persistent cough and wheeze, unresponsive to salbutamol.

He has mild respiratory distress on occasion but has never required hospital admission. He was born at 27 weeks' gestation and had a difficult first 2 weeks of life: he was ventilated for respiratory distress syndrome and had chronic lung disease. At first I thought he had bronchiolitis but this has persisted for a while and his mother is anxious.

He has a developmental follow-up clinic at City Hospital in 4 months but I would be grateful if you could see him before this. His mother has attended my surgery four times in the last 2 months.

Yours sincerely,

Dr Jefferies

General Practitioner

Background information

Weight and height on 9th centile.

Other information

Mother is a respiratory nurse on an adult ward in this hospital.

Preparation time (2 min): initial thoughts

You are going to need to take a detailed history of Samuel's symptoms as well a a neonatal history. His mother may have some ideas about what she thinks th problem might be.

Consultation

Introduction. Clarify role, agree aims and objectives.

History

- ○ Current symptoms – persistent or intermittent wheeze? Any days wher there has not been a problem?
- ○ Did the problem start suddenly or gradually?
- ○ Associations with coughs, colds, etc
- ○ Presence or absence of fevers, other features indicating that illness is affecting the infant such as poor feeding, weight loss
- ○ *Thorough birth and neonatal history – a template is given below for any cas involving an ex-premature baby; it needs to be adapted for the situation:*
    - Details of pregnancy (including maternal health, smoking)
    - Delivery/resuscitation
    - Respiratory management on neonatal unit and number of days in oxygen
    - Was home oxygen required?
    - Go through a 'checklist' of other potential problems on the neonatal unit (NNU), eg patent ductus arteriosus, interventricular haemorrhage, retinopathy of prematurity, feeding problems (length of time on total parenteral nutrition (TPN) if relevant), jaundice, sepsis.
    - Total length of time on NNU
    - Feeding history. Are there reflux symptoms?
    - Other medical problems/admissions. Always ask about eczema and food allergy and inhalation of foreign bodies in wheezy infants
    - Developmental history (detail depends on the case)
    - Medications (current and previous) and immunisations
    - Consultant in charge/follow-up previously arranged.

Make sure you address the respiratory problems in detail and do not explore these other issues to the detriment of finding out the cause of the wheeze.

Warning bell rings; you have 4 min left with Samuel and his mother. You have gathered the following information so far:

Samuel was well until he developed a runny nose followed by a cough and mild respiratory distress 2 months ago. Hospital admission was considered but his saturations were 97%, feeding was minimally affected and his mother was fairly confident she could manage him at home. His GP thought he had bronchiolitis. This cough and wheeze persisted for 3 weeks and the GP was visited a second time. Salbutamol was tried at this stage but did not work. There was then a period of about a week where symptoms subsided but then he started wheezing again. Again, this lasted 2–3 weeks and he gradually got better. In the past week it has happened again and his mother has visited the GP twice to discuss other treatments. She is concerned it may be aspiration secondary to gastro-oesophageal reflux but no treatments have been started yet. You have asked about symptoms of reflux; there do not seem to be any present. There has never been any stridor.

Samuel is thriving.

He was born at 27 weeks after his mother had an antepartum haemorrhage. He needed 1 week of ventilation and 1 week on nCPAP. He was on oxygen for nearly all of his 15-week admission.

Inotropes were required for the first 2 days. He was transfused. There were no episodes of sepsis. He was on parenteral nutrition for 10 days and was given breast milk from day 2. Development is normal for corrected gestation.

Remaining 4 min:

You take a social history. There are no major issues; she and her husband also have a 4-year-old boy who has just started nursery. You also try to explore her concerns and give her some time to air them.

She still feels that reflux might be a reason for these episodes of wheeze and is worried that his lungs will not recover from all the 'previous damage'. You verify the main points and summarise the history. There are no further questions from Samuel's mother.

The bell rings and you are left with the examiner.

Discussion with examiner (9 min)

Would you summarise this case for me?

I saw Samuel, a 14-month-old boy born at 27 weeks' gestation so corrected to 11 months. His main problem on the neonatal unit was respiratory distress syndrome (RDS) and he developed chronic lung disease. His current problem is recurrent cough and wheeze, which started after an episode of bronchiolitis. He has had two other viral illnesses since then so has been wheezy a lot of the time.

What would you say to his mother ?

A first episode of bronchiolitis can leave a residual wheeze in any patient that can take a while to settle. This is especially true of ex-premature babies. It does not often respond to ipratropium bromide or salbutamol, but both are worth a trial. The most likely cause of the recurrent episodes of cough and flare-ups of wheeze (especially with the brother in nursery) is two or three separate viral lower respiratory tract infections (LRTIs) affecting small, hyper-reactive airways. Inhaled steroids are not usually warranted for this situation. I would advise that her child is seen by a doctor if he is acutely unwell with the wheeze or is failing to gain weight adequately.

What other causes would you consider in a persistently wheezy infant or toddler and how would you investigate them?

- ❍ Inhaled foreign body – inspiratory and expiratory chest X-ray
- ❍ Pertussis – pernasal swab
- ❍ Multi-trigger wheeze/true early asthma – demonstrable reversibility of symptoms, atopy, family history
- ❍ Cystic fibrosis – sweat test
- ❍ Congenital airway malformation – bronchoscopy
- ❍ Early asthma (toddler).

Notes on problems faced by ex-premature babies

Families whose children were premature and who were on the neonatal unit for a long stay are often happy to help out in exams. It is therefore useful to brush up on all of the common problems that ex-premature babies may have faced and in particular which of these may still be affecting them. It is useful to attend your nearest neonatal follow-up clinic to see these issues first hand, especially if it has been a while since your last neonatal job.

You should be able to discuss the following with examiners and indeed parents, as these topics may also crop up in a communication scenario. They can all be found in standard neonatal texts or review articles.

Chronic lung disease

The definition of chronic lung disease states that the child must have required respiratory support with or without ventilation after birth and still be oxygen dependent at 28 days.

Prognosis is a common parental concern. Most babies with chronic lung disease can be weaned off oxygen before discharge. The rest need home oxygen setting up (a Home Oxygen Order Form needs to be filled out), which can delay discharge if not planned well in advance. Weaning is then undertaken slowly, usually within a few weeks or months. Many patients have ongoing problems with reactive airways. They are especially symptomatic during episodes of bronchiolitis; the illness can be more severe and the effects can last for longer than in full-term babies. Thresholds for admission should therefore be lower. Most babies with chronic lung disease require at least one admission per year for the 2 years post discharge. Do mention the importance of immunisation against RSV and an annual influenza vaccine in these infants.

Patent ductus arteriosus

Treatment is supportive, but medical therapy may well have been required. This is with indomethacin or ibuprofen. Remember the risks associated with indomethacin (ie renal impairment and necrotising enterocolitis). Ibuprofen is increasingly being used, as it is just as efficacious and has fewer renal side-effects.

Surgical therapy (duct ligation) may also have been required. This is usually considered when a baby has been unresponsive to two rounds of medical therapy, has a large duct on echocardiography and/or is still ventilator dependent.

Feeding/nutrition

Parenteral nutrition (PN) and its complications (extravasation injury, infection/vascular thrombosis secondary to long lines, conjugated hyperbilirubinaemia) are often a source of concern. Difficulties in establishing feeds, poor weight gain and necrotising enterocolitis are other problems that may have been experienced by ex-premature babies.

Germinal matrix haemorrhage (GMH) and intraventricular haemorrhage (IVH)

Premature babies (especially those under 32 weeks) are particularly prone to intracranial bleeds. These very often start in the subependymal germinal matrix, which is a highly vascular area. Bleeds are graded on ultrasound, and can cause further problems:

- ○ A germinal matrix haemorrhage can extend into the ventricular space, in which case it is termed an intraventricular haemorrhage. This can lead to progressive ventricular enlargement, which needs to be closely monitored by measuring the Ventricular Index on ultrasound. On the severe end of this spectrum is post-haemorrhagic ventricular dilatation (PHVD) which can lead to hydrocephalus, and the baby may require a ventriculo-peritoneal shunt.
- ○ Extensions of IVH into brain parenchyma can later result in the formation of cysts.

The incidence of IVH has decreased as a result of general improvements in neonatal care.

You may be asked about the prognosis. Babies with only a GMH or a small IVH have a good outcome. At the other end of the spectrum, those with a large IVH have a 50% chance of developing significant neurodisability.

Periventricular leukomalacia (PVL)

This is the most common brain injury sustained by preterm infants, and stems from multifactorial insults (namely inflammatory, hypoxic, ischaemic) to the periventricular white matter. The end result is loss of white matter, with or without cystic changes.

There is often a resulting motor impairment (descending corticospinal tracts), leading to cerebral palsy which is variable in severity. Cortical blindness is another risk.

Visual problems

May be due to retinopathy of prematurity (ROP), PVL or to direct cortical damage. If the baby had ROP, ask if laser treatment was required. This may prevent blindness from retinal detachment but can also cause peripheral field defects.

Squint is also commoner in ex-premature babies than those born at term.

Patients with any visual problems are followed up in neonatal and ophthalmology clinics.

Hearing problems

All ex-premature babies should have had a hearing test. If hearing is suboptimal ask about risk factors, ie aminoglycosides, jaundice, hypoxia, IVH and PVL. They will need follow-up, usually by a community paediatrician with a special interest in audiology or a consultant paediatric audiologist.

Developmental delay and learning difficulties

Although in some cases this may not be a major problem when corrected for prematurity, it is a huge concern for parents. There is a higher than normal incidence of learning difficulties even in more mature ex-preterms (eg 32 weeks' gestation), who may appear normal at 2 years of age. A smooth transition between the neonatal follow-up clinic and child development clinics is required for infants whose development is a cause for concern.

2.4 Case four: recurrent abdominal pain

Role

You are the Specialist Trainee.

Setting

Outpatient department.

You are talking to

Mrs Oliver and her 9-year-old daughter, Milly.

Dear Dr...

Re: Milly Oliver

Thank you for seeing Milly, who is 9 years old and has complained of central abdominal pain on and off for the last 4 months. Her attacks are painful and she has been sent home from school on numerous occasions. Her brother Saul has constipation and I saw him 2 weeks ago. Mrs Oliver says that she has no concerns about Milly's bowel habits but is concerned at the amount of school she's missing. Examination today was normal. Although an organic cause is unlikely, I would be grateful if you could assess her to see if this needs investigating further.

Yours sincerely,

Dr Lincoln

General Practitioner

Background information

Weight and height on 25th centile.

Preparation time (2 min): initial thoughts

You will need to focus your history very well; this may avoid unnecessary investigations. A careful history including social history and an enquiry around ideas, concerns and expectations is particularly important.

Consultation

Introduction. Clarify role, agree aims and objectives.

History

As with all cases, ask open-ended questions and listen. Explore and note the psychosocial elements of the illness as you go. Always try to build up a picture of how the illness affects the child's daily living:

○ When did the pain start? Any obvious triggers?
○ Location, intensity, character and duration, time of day or night
○ Does pain wake her at night?
○ Exacerbating and relieving factors
○ Appetite, diet, satiety
○ Presence of nausea, emesis, reflux symptoms, urinary symptoms
○ Stool pattern, consistency, completeness of evacuation
○ Weight loss, growth delay, rash, joint pains or eye symptoms
○ Past medical history, focusing on relevant previous illnesses and gastrointestinal problems
○ Ask about previous health professionals involved with this problem, and their views/explanations (may differ from yours)
○ Drug history, allergies (especially food allergies)
○ Family history of migraine, irritable bowel syndrome, inflammatory bowel disease
○ Social history (much may have been revealed already): school (ask about absence), hobbies/play (how badly affected?), peer relationships, family dynamics.

Warning bell rings; you have 4 min left with Milly and her mother. You have gathered the following information so far:

Milly began to get the pain over 4 months ago. This seemed to come 'out of nowhere'. Now occurs on most days and can last from 20 minutes to 2 hours. Paracetamol helps. She feels it 'near her belly button'. She does feel sick but has not vomited. She occasionally looks pale with these attacks. Her appetite is occasionally reduced. Bowels open once or twice daily and normal consistency according to her mother. Past medical history unremarkable, not on medication. Brother has constipation, mother has migraine.

Remaining 4 min:

You take a social history and learn that Milly lives with her parents and brother. There do not seem to be any problems at school but she is a shy girl and 'a bit of a worrier'. Her father is in the army and is away for half the year. You ask Milly and her mother if there are any other concerns and are told that she has missed about 2 weeks' worth of school in the past 2 months. Mrs Oliver is very concerned that this is Crohn's disease, which her sister (Milly's aunt) has been battling with for 4 years.

The bell rings and you are left with the examiner.

Discussion with examiner (9 min)

Would you summarise this case for me?

I saw Milly, who is 9 years old, with her mother. She has been suffering from recurrent bouts of central abdominal pain that have resulted in her missing 2 weeks' worth of school in the last 2 months. She has not lost weight and has no obvious pointers to an organic cause in the history I obtained. There is however a family history of Crohn's disease, but not in a first-degree relative.

What are pointers towards a pathological cause?

Warning signs in chronic abdominal pain are:

- Very young child
- Pain well localised and further out from the umbilicus
- Vomiting (especially bilious)
- Unexplained fever
- Change in bowel habit especially diarrhoea and/or blood in stool
- Nocturnal waking with pain
- Weight loss
- Poor growth
- Family history of coeliac disease or inflammatory bowel disease (especially in first-degree relatives)

What would your initial management be in this case?

Cover the following:

- In this case *some* investigations to exclude commoner organic causes (especially as family history of inflammatory bowel disease) would be justifiable:
  - Full blood count, inflammatory markers, liver function tests, coeliac screen, stool culture, urine for microscopy, culture and sensitivities (MC&S). An abdominal ultrasound scan is worth considering, and if normal could also play a part in reassuring the child and family.

- Encourage the child to keep a symptom diary (including any dietary, psychological or social triggers) that can be reviewed and reflected upon with the child and family at follow-up. If investigations are normal and no pathological diagnosis is apparent, then reassure that there is no serious pathology. Acknowledge parental concern about any family history of GI problems, and reassure when appropriate.
- An explanation that the most likely cause is functional abdominal pain (followed by an explanation of this term) is then reasonable. Show sympathy toward the child and family and acknowledge that the pain is very real, despite the absence of a demonstrable physical cause. Explain how pain can occur in the absence of disease. Advise that the child should try to attend school as much as possible and if appropriate, advise regarding school reintegration.
- Simple analgesia when pain first comes on, while avoiding excessive medication.

What if these strategies do not work?

- Referral to the Child and Adolescent Mental Health Team/psychologist may be necessary in severe cases, particularly if there are concerns about the child's mental health or if there is evidence of disordered eating/anxiety around food. Such teams can look further into family dynamics and hopefully help with non-pharmacological pain management techniques
- Referral to paediatric pain team (can often help minimise analgesia; some patients take excessive or inappropriate analgesics)
- Liaise with school (with parental consent) to explore any obvious barriers to school attendance.

Notes on recurrent abdominal pain (RAP)

This is abdominal pain, significant enough to interfere with normal activity, that has occurred more than twice in a 3-month period. It is relatively common in younger school children and adolescents. Children with recurrent abdominal pain may be invited to the exam for a session, to reflect the kind of patients that you will meet when you conduct outpatient clinics more regularly.

The majority of patients with RAP have *functional abdominal pain.* This means that there is no evidence of a pathological condition causing the pain. It is now believed that children with functional abdominal pain have abnormal bowel reactivity to physiologically and psychologically stressful stimuli.

Four categories of functional abdominal pain are described as part of the Rome III criteria:

- Functional dyspepsia – upper abdominal pain often related temporally to food

- Irritable bowel syndrome – alternates between episodes of loose stools and constipation
- Abdominal migraine – attacks involve pallor, nausea and sometimes vomiting in a child with a family (especially maternal) history of migraines
- Functional abdominal pain – none of the above features but other causes excluded.

General management has been outlined above; it is important to realise that functional abdominal pain is by definition a diagnosis of exclusion and that some laboratory investigations and a degree of follow-up/ongoing support will often be necessary. Family education and their acceptance of the diagnosis is very important and a positive prognostic factor.

It is important to recognise that functional symptoms can occur in children with an underlying organic disease, so safety-netting is an important part of management.

In children with functional abdominal pain, practical management involves coping strategies and rehabilitation. It is important that the child returns to normal functioning, even if the pain cannot be completely eradicated. Input from a psychologist may be needed in difficult cases. Pharmacological treatment is generally not helpful unless there are specific problems such as constipation. Lifestyle strategies including healthy eating, plenty of fluids and regular exercise are generally helpful.

2.5 Case five: cerebral palsy

Role

You are the Specialist Trainee.

Setting

Outpatient department.

You are talking to

Mrs and Mrs Melling and their 9-year-old son, Ashley.

Dear Dr…

Re: Ashley M

Thank you for seeing this 9-year-old boy who has quadriplegic cerebral palsy. He has moved here from New Zealand recently and the family have come to see me to get him registered. His mother has a clinic letter from 6 months ago detailing his main problems. They are most concerned that his hips are getting more painful with significant spasm. His scoliosis is much more pronounced than it was a year ago. They could not attend his final appointment before they left for the UK. I thought it best that you meet up with the family soon. I would be grateful if you could also refer him to the correct agencies to continue/add to the multidisciplinary care provided in New Zealand.

Yours sincerely,

Dr S Smith

General Practitioner

Background information

Parents have forgotten the letter from New Zealand.

Additional information

Medications – sodium valproate 250 mg bd.

Preparation time (2 min): initial thoughts

You are not going to be able to take a complete, exhaustive history in the 13 minutes available. You are being asked to address the main problems (particularly contractures and scoliosis) in order to refer him to the most suitable agencies.

Consultation

Introduction, clarify role. Agree aims and objectives.

Remember to include the child in the consultation and make good eye contact, even if there seems to be little reciprocation. Many children with cerebral palsy are able to understand you; do not assume that no children with this problem have normal intelligence.

History

❍ When was the news first broken that Craig has cerebral palsy? How was it diagnosed and by whom? If there were obvious problems from birth, start with prenatal history
❍ Results of previous investigations if relevant, eg neuroimaging. This is especially important to ask as there may be a known cause, such as polymicrogyria
❍ Major medical problems – neurological, feeding and weight, musculoskeletal. Which problem is the biggest for parents and which for the child? This is a good question to ask when there is a multitude of problems and little time to discuss them all. Try to get an idea of the child's quality of life
❍ Professionals involved so far (try to link with problems, eg feeding = dietician, speech therapist, paediatrician)
❍ Previous management of contractures – professionals involved, especially physiotherapist (*in this case it is worth spending longer on this problem)*
❍ Birth history (if not already taken) – with special attention to any problems antenatally
❍ Developmental history, if there is time
❍ Medications
❍ Social and family history
❍ Then ask, 'Is there anything else we haven't covered that I should be aware of ?'

Warning bell rings; you have 4 min left with Ashley and his parents. You have gathered the following information so far:

Ashley had an extremely difficult start to life. He was born by emergency caesarean section at 42 weeks following a prolonged bradycardia. His parents recall that he had cardiac massage after birth. He was initially ventilated and had significant neonatal seizures. He never managed to feed properly and had a nasogastric tube, which the parents learned to use at home when he was eventually discharged.

He had an MRI scan of his head as a baby, which showed some 'significant damage'.

His main problems are severe developmental delay, gastro-oesophageal reflux and poor weight gain (for which he had a gastrostomy and fundoplication) as well as progressive hip dislocation and scoliosis.

He has epilepsy, which is well controlled. He is wheelchair-bound. Ashley can communicate using Makaton. His vision and hearing are normal.

His parents feel that 'his hip pains are bothering him most'. He was being seen regularly by a physiotherapist and orthopaedic surgeon for this problem, and he was being considered for hip surgery before the family moved.

Ashley has also been seen previously by a general paediatrician, community paediatrician, paediatric surgeon, dietician and occupational therapist.

In the remaining 4 min you try to focus on schooling and family life.

The family have already approached several schools and they will need written reports from the paediatric/community teams that will go towards a needs assessment.

Their new home is a large ground-floor flat and they are already looking at ways to modify one of the rooms. Ashley is an only child. His parents are setting up a business in the UK and are currently working from home. They have savings that mainly go towards Ashley's care.

The bell rings and you are left with the examiner.

Discussion with examiner (9 min)

Would you tell me what the main issues are for Ashley?

Ashley is a 9-year-old boy with quadriplegic cerebral palsy whose family have just moved to the country. His main problems at the moment are painful, dislocated hips and his progressive scoliosis. His other problems are epilepsy, severe learning difficulties and poor weight gain. On taking a social history, I feel that the family are not yet fully settled in the UK.

What are the different ways in which you could help Ashley and his parents settle into their new country?

There are several options:

○ Ashley needs to be referred to the local child development centre/community paediatric team so that his different care needs can be best met.

- He should be referred to a paediatric orthopaedic surgeon and a spinal surgeon as well as a physiotherapist. An occupational therapist could help to install suitable equipment in the new flat.
- His epilepsy would need to be regularly reviewed and he should be referred to an epilepsy nurse.
- Ongoing care can be facilitated by conducting a multidisciplinary team meeting so that the parents can meet the professionals involved and agree upon an agenda, including time schedules for the various aspects of care. A multidisciplinary needs assessment will help ensure that he attends a suitable school.
- Financial – support application for Disability Living Allowance (DLA) and any other relevant benefits.
- Giving his parents contact numbers for support groups in the area, and organisations such as Contact a Family.
- Arranging some local respite care so that parents can have a break, especially as they are new to the country.

Why has Ashley got such severe feeding problems?

Feeding problems in children with cerebral palsy are multifactorial and many factors are potentially implicated. Pseudo-bulbar palsy is likely to be a major factor and this is present in many children with quadriplegic cerebral palsy. This will lead to difficulty swallowing and potentially primary aspiration. He could also have residual gastro-oesophageal reflux even after fundoplication. There are likely to be other factors including mobility, posture, seating, spinal disease, constipation, stress, etc.

Notes on management of cerebral palsy

The cerebral palsies can be defined as persistent disorders of movement and posture due to a non-progressive disorder of the developing immature brain. Motor function is not simply delayed, but also follows a course that is not seen in the normal child.

There are often accompanying problems with other aspects of development such as sensory, social and cognitive.

This definition excludes diagnoses such as brain tumours and spinal cord disease.

Prevalence is around 2 per 1000 children.

Diagnosis

Cause unknown in many, and many factors can be responsible. Unless the cause is obvious, try to elicit/note some of the following problems/risk factors in the history:

Prenatal

- ○ Genetic forms – autosomal recessive and autosomal dominant
- ○ Cerebral malformation
- ○ Maternal alcohol consumption/substance abuse
- ○ Infection – *Toxoplasma*, rubella, cytomegalovirus, HIV, UTI in pregnancy
- ○ Intrauterine growth retardation, small or large for gestational age infant
- ○ Prematurity, particularly under 32 weeks, and the risk increases with decreasing gestational age at delivery
- ○ Multiple pregnancy.

Perinatal

- ○ Neonatal encephalopathy – if so, ask about seizures; there may have been neuroprotection (cooling)
- ○ Ventricular haemorrhage (ask about head scans)
- ○ Episodes of hypoglycaemia: severity and duration.

Postnatal

- ○ Meningitis/encephalitis
- ○ Head injury
- ○ Stroke.

Investigation

You may be asked how to (or whether you would) investigate a child with cerebral palsy. This is a rapidly changing field particularly with the increased availability of genetic testing and it would be sensible to read a review/discuss with your local paediatric neurodevelopmental/neurology team the most up-to-date guidance.

Investigations are performed for several reasons, unique to each child. Looking for an alternative diagnosis is one reason, especially when:

- ○ No obvious cause can be found after a comprehensive history has been taken
- ○ There has been a loss of skills
- ○ Familial occurrence of a similar neurological problem
- ○ Encephalopathy
- ○ Unusual features, (eg ataxia, nystagmus)
- ○ Lower motor neurone signs
- ○ Symmetrical signs

Useful investigations include:

- CT or MRI of head – MRI in most cases, especially if neuronal migration defects are being considered
- MRI of spine if a cord lesion is suspected
- Chromosome analysis – microrray-based comparative genomic hybridisation ('array CGH')
- Electroencephalogram (EEG)
- Congenital infection screen
- Metabolic screen, including blood, urine and CSF studies
- Ophthalmological assessment
- Hearing tests.

Different patterns of cerebral palsy and their specific problems

Motor involvement can be described in terms of whether it is spastic or dystonic (can be both), symmetrical or asymmetrical. Describe the limbs involved and then whether the face is involved. Disorders of tone/movement are then described where relevant.

Hemiplegia and diplegia

In hemiplegia there can be varying degrees of spasticity in the affected side of the body, requiring physiotherapy to prevent the worst deformities from developing. Weakness is usually more pronounced distally. The incidence of epilepsy and related disorders is related to cortical involvement. Sensory defects, visual field defects and cranial nerve involvement can all occur. Most children will walk by 18–24 months; if not, other pathologies should be considered or the cerebral palsy extent reviewed.

In *diplegia* the limbs on both sides of the body are affected, legs more than arms. More severely affected children with lower limb involvement never walk. It is more common in preterm low-birth-weight infants and in those who had an abnormal labour or delivery.

Quadriplegia

There is four-limb involvement, with the arms more severely affected than the legs. Initially, young children are hypotonic with episodes of axial stiffening and extension. As they grow older the tone increases, producing rigidity with pyramidal signs. Spasticity and contractures can be severe and often need medical and sometimes surgical management. The 'windswept' hip deformity is particularly characteristic, the adducting hip being at risk of dislocation. Scoliosis can be severe and should be treated aggressively.

Bulbar muscles, usually spared in hemiplegia and diplegia, are commonly affected. Severe learning difficulties and epilepsy are common. Many affected infants are microcephalic.

Athetoid cerebral palsy

This involves involuntary movement of the limbs. Choreoathetosis is largely associated with kernicterus (secondary to neonatal hyperbilirubinaemia) or a neonatal encephalopathy.

Ataxic

Here the motor disorder is cerebellar ataxia, but this is usually associated with diplegia. It is usually diagnosed relatively late – patients need to reach a certain stage of development before cerebellar deficit is noticeable. It is difficult to diagnose and you must look for other causes of cerebellar dysfunction, eg tumours and other progressive disorders (Friedreich's ataxia, ataxia telangiectasia, metachromatic leukodystrophy).

There are some children who fall into the category of mixed forms of cerebral palsy, eg ataxia and diplegia.

General problems encountered in cerebral palsy

The treatment of problems that arise in cerebral palsy is something you may be expected to talk about at length.

Remember that while an appreciation of all of the different problems encountered by children with cerebral palsy is important, it is crucial to be able to determine the overall functional ability of the child, and to work with that.

You should try to describe and discuss each patient in terms of the following:

- Spasticity. This can be a huge problem, causing pain, additional functional impairment and eventually contractures. It can be treated with baclofen and in some cases botulinum toxin, which is also used for medical treatment of well-established contractures. Splints may be used at night to try to prevent the onset of contractures, and special shoes can help to correct unequal leg length that may result from limb shortening. Orthopaedic surgery has much to offer (eg elongation of the Achilles tendon), although this is rarely required before 5 years of age. Selective dorsal rhizotomy is another procedure that may be considered.
- Dystonia
- Baseline level of function (this is usually documented by members of the child development centre team, including physiotherapists and occupational therapists) including ability to communicate with those around them

- Medical co-morbidities (eg nutritional status, gastro-oesophageal reflux, constipation, seizures, multiple chest infections)
- The wider social implications for the child and their family.

Management – professionals involved

Management involves a *multidisciplinary approach*, usually with the involvement of a child development unit. The team will comprise:

- **Community paediatrician:**
  - Usually in a co-ordinating role; patients may see a general paediatrician as well as a community paediatrician
- **Physiotherapist:**
  - Has a fundamental and crucial role in assessment and the planning of therapy
  - Is involved with the child from an early stage
  - Helps in preventing severe contractures and deformity as well as aiding as much normal motor development as possible
- **Occupational therapist:**
  - Works closely with physiotherapist and is involved in adapting the environment to aid the child's functioning
  - Is involved in adapting play within the home to aid development
- **Communication therapist:**
  - Is involved early, providing help with feeding problems
  - Encourages parents to stimulate their children, and to help them develop as much communication as possible, whether it be normal speech, sign language or a mixture of both
- **Social worker:**
  - Works alongside all the therapists that have close links with the family
  - Provides support for the family, helping with periods of stress
  - Arranges holidays and social admissions/respite care should the need arise
- **Others include:**
  - Dietician; educational psychologist; psychologist; play therapist; teacher; ophthalmologist; orthoptist; audiologist; respiratory paediatric team; paediatric neurology team; ear, nose and throat (ENT) surgeon; orthopaedic surgeon.

2.6 Case six: nephrotic syndrome – relapse

Role

You are the Specialist Trainee.

Setting

Paediatric assessment unit.

You are talking to

Mr Watts and his 8-year-old son, James.

> Dear Dr…
>
> Re: James Watts
>
> Thank you for seeing this 6-year-old boy who had nephrotic syndrome diagnosed 2 years ago. He was given a treatment course of prednisolone and required no other medications. He was not referred to a nephrologist. In the past 4 days he has had mildly puffy eyelids and 2+ of protein in his urine. He had just got over a cold. I am concerned that this is a relapse and would be grateful if you could review him.
>
> Yours sincerely,
>
> Dr. Black
>
> General Practitioner

Background information

Weight tracking 25th centile, systolic blood pressure 'high' at GP surgery.

Preparation time (2 min): initial thoughts

You will need to focus on his previous illness and start thinking about how the current episode should be managed.

History

○ Introduction, clarify role. Agree aims and objectives. Remember to introduce yourself to and engage with the child.

- ❍ When were parents first concerned this might be a relapse?
- ❍ Any other symptoms apart from puffy eyelids?
- ❍ Ask about general health at the moment.

Confirm and discuss previous diagnosis

- ❍ How did he initially present?
- ❍ Medications used to treat and days before achieved remission
- ❍ Rule out previous complications such as difficulties in fluid management (hypovolaemia), infection, blood clots (thrombosis)
- ❍ Ask if there are any other features that concerned doctors, such as high blood pressure, renal impairment, blood in urine (macroscopic haematuria). Although he was not referred to/did not see a nephrologist, he may have been *discussed* with one
- ❍ Maximum weight if remembered
- ❍ Ask if he is still under paediatric follow-up
- ❍ Ask about other illnesses in the past (ask specifically if he has had chickenpox)
- ❍ Medications
- ❍ Immunisations (ask specifically if he has had pneumococcal vaccine and influenza vaccine)
- ❍ Family history of renal disease or any vasculitic illnesses such as systemic lupus erythematosus (SLE).

Bell rings for 4 min. You have obtained the following information:

James was diagnosed 2 years ago. He initially had puffy eyes for 5 days. This was thought to be due to hayfever and he was given an antihistamine. However, this swelling did not go down and his face then started to look swollen. He had put on half a kilogram in this time and so was referred to hospital. His urine dipstick test showed +++ of protein. Some other blood tests were taken and he was started on prednisolone daily. He was not given any other medications apart from penicillin. For a couple of days he produced less urine but his kidney function on the blood tests was normal and 'his protein levels were a bit low, but he didn't need any more'. After 16 days his urine dipstick was negative for protein.

He has been well since but developed a cold a week ago. The family had been told to dip his urine in the morning when unwell and this became positive for protein 3 days ago. His eyes look a bit puffy again.

You remember to obtain a family history, which is unremarkable for any renal problems. James lives with his parents and is an only child.

You ask the father if he has any other concerns; he is worried that 'this will happen again and again'.

The bell rings and you are left with the examiner.

Discussion with examiner (9 min)

Tell me about this young man's problems

James was diagnosed with nephrotic syndrome 2 years ago and was treated with prednisolone. He went into remission after 16 days and had no complications. He now presents with a relapse of his nephrotic syndrome and has had a high blood pressure recorded at the GP's surgery.

What is your definition of a relapse of nephrotic syndrome and how common is this?

Relapse is defined as a protein of 2+ or more on dipstick for 3 days consecutively. Only one-third of patients will never have another relapse. Relapses often follow infections and parents should be told to dip their child's urine every morning if they are unwell.

Would you treat this child?

Yes, as he has had 4 days of proteinuria. I would start prednisolone 60 mg/m^2 per day until remission, along with penicillin prophylaxis.

When would you discuss him with a paediatric nephrologist?

When there are any atypical features (some of which would be indications for renal biopsy):

○ Age < 1 year, > 12 years
○ Renal impairment
○ Persistent hypertension
○ Reduced C3
○ Gross haematuria
○ Systemic features of a vasculitis such as rash, arthropathy.

And/or under the following circumstances:

○ There are any acute complications such as severe hypovolaemia, infection or thrombosis
○ Not in remission by day 28 of prednisolone (steroid resistance)
○ Two consecutive relapses on steroid treatment or relapse within 14 days of stopping steroids (steroid dependence)
○ Frequent relapses.

At this moment in time I would not discuss him with a nephrologist but would review this decision dependent on his progress.

But what about his blood pressure? It was raised at the GP's surgery

Needs repeating on three occasions with the correct cuff size. Look up blood pressure for height centile, age and sex on centile charts. If still over 95th centile, discuss with a nephrologist. May in the acute setting reflect hypovolaemia.

Notes on nephrotic syndrome

This is a triad of:

- Proteinuria (early morning protein:creatinine > 200 mg/mmol)
- Oedema
- Hypoalbuminaemia (< 25 g/l)

Incidence in the UK is around 2–4/100 000, with boys being more commonly affected than girls. Presentation is usually between 18 months and 6 years of age and is sometimes confused for an allergic reaction or idiopathic angioedema, as the oedema is initially periorbital. The oedema then becomes dependent.

Children with nephrotic syndrome are often asked to participate in the exam. They can be brought up to the exam whether they are currently suffering from symptoms or not. It is worth having a thorough knowledge of this condition as there are many aspects of the management that can help examiners discriminate between well-prepared and less well-prepared candidates.

Investigations

These are undertaken to confirm the diagnosis and also to look for any atypical features and complications:

- Urine dipstick, MC&S, early morning protein:creatinine ratio, urinary sodium
- Blood for full blood count (FBC), urea and electrolytes (U&Es), albumin, cholesterol, C3 and C4, varicella zoster titres, hepatitis B and C serology
- If systemic or atypical features are present as outlined above, then further autoimmune profile including antinuclear antibody and anti-double-stranded DNA.

General management

- Monitor and control fluid balance – need input/output charts, daily weights. Some patients need fluid restriction and diuretics. Need to watch for hypertension
- Dietician involvement – needs low-salt, normal-protein diet
- Look for and manage complications (see below), particularly hypovolaemia.

Pharmacological management

Steroids are the mainstay of management in most cases. In Europe around 90% of children with nephrotic syndrome have steroid-sensitive nephrotic syndrome (SSNS). These patients respond to initial high-dose prednisolone (60 mg/m^2) for 4 weeks, followed by 40 mg/m^2 on alternate days for 4 weeks, then cessation.

If the patient is not in remission by day 28, they are said to have *steroid-resistant* nephrotic syndrome (SRNS). Renal biopsy will need to be considered. Second-line treatment agents such as cyclophosphamide, cyclosporin, tacrolumus or levamisole are used.

Relapses are treated with a daily dose of steroid, with a switch to an alternate-day regimen as soon as remission is achieved.

Some children respond to steroids but relapse while weaning or in less than 2 weeks from stopping treatment. This is termed *steroid-dependent* nephrotic syndrome.

A child is said to have *frequently relapsing* nephrotic syndrome if they have two or more relapses in the first 6 months or over four relapses within a year.

These children are also considered for second-line treatments but in cases of multiple treatment failure or intolerance, other treatments such as mycophenolate mofetil (MMF) or Rituximab, a monoclonal antibody, can be used.

It is worth familiarising yourself with your local renal unit protocol for this condition.

Complications you may need to discuss

Poor knowledge of these is a good discriminator in exams.

Hypovolaemia

Reduced capillary oncotic pressure (secondary to protein leak from the kidneys) causes an efflux of plasma water from the circulation. Eventually, homeostatic mechanisms to conserve vascular volume cannot keep up and the patient becomes symptomatic.

Clinical features of hypovolaemic shock are abdominal pain (splanchnic ischaemia), tachycardia, prolonged capillary refill time, wide core–peripheral temperature gap, and decreased urine output.

Urine sodium will be low with a compensatory rise in potassium.

Treatment is with fluid resuscitation with sensible amounts of isotonic saline, eg 10–20 ml/kg, then assessment of clinical response. After discussion with the renal team, patient may need 5% or 20% albumin for more sustained intravascular volume expansion. Be aware that 20% albumin in particular can cause rapid refilling of vasculature and resultant left ventricular failure with severe pulmonary oedema.

Infection

Patients are at increased risk of infection due to urinary loss of immunoglobulin and complement. They are particularly prone to pneumococcal and *Haemophilus influenzae* infections, particularly peritonitis and septicaemia. Prophylactic penicillin should always be given while there is proteinuria. Pneumococcal and influenza vaccines are recommended.

High-dose steroids may mask signs of infection and there can be diagnostic confusion with hypovolaemia, especially if patient is peritonitic, as both cause abdominal pain and circulatory compromise.

A suspicion of peritonitis warrants broad-spectrum antibiotics and a surgical opinion.

Nephrotic children are also vulnerable to chickenpox (needs aggressive treatment with aciclovir and varicella zoster immunoglobulin if there are lesions) and measles. They are also defined as being immunocompromised from their steroids if they have had > 1 weeks' worth of steroids in the past 3 months or any other form of immunosuppression in the last 3 months. In these cases they should have been given a steroid card. Live vaccines should not be given until the child is off steroids for 6 months.

Thrombosis

Due to renal loss of antithrombin-3 and hyperviscous blood. Renal vein thrombosis can occur, and can present with macroscopic haematuria. Thrombosis is less of a risk if patients are adequately hydrated and mobilisation encouraged.

Parental education

This is an important part of management. Parents need to be given written information on the illness and should be aware of the complications. In particular they should be told to contact the ward urgently if their child develops abdominal discomfort. They should have a written regimen for tapering the steroids at home and be aware of side-effects (a common question in the Communication Station). Urine should be dipped daily until remission, then twice weekly while still on a tapering dose of steroid or if their child is unwell at any time. Results can be recorded in a diary.

3

FURTHER SCENARIOS

The six detailed examples in Chapter 2 demonstrate the depth of knowledge required when preparing for the examination, and some of the different types of history taking scenarios that you might meet.

This chapter contains some additional suggested topics for practice scenarios. Use the framework for approaching this station as provided and go through the examples with your colleagues. The 12 examples here (along with the six cases in Chapter 2) do not constitute a syllabus for the examination but once you are comfortable talking through the main problems associated with this broad range of illnesses/scenarios, you will feel better armed for the station. Many of the conditions mentioned here are covered in other sections of this book. The information that follows each scenario is not exhaustive, but has been adapted for this station and reflects the *questions you shouldn't miss asking* as well as *what you may be asked by the examiners*. Much of this knowledge base has been acquired from clinical experience, and also from centralised guidelines such as those from the Royal College of Paediatrics and Child Health (RCPCH), National Institute for Health and Care Excellence (NICE) and the Scottish Intercollegiate Guideline Network (SIGN) that are available and should be reviewed by the candidate.

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3.12 Scenario twelve: Review of patient with extrahepatic biliary atresia post liver transplant

3.1 Scenario one: coeliac disease in a 4-year-old boy; 1st follow-up appointment

Presented initially with fatigue, then abdominal pain and distension, chronic diarrhoea and faltering growth. Confirmed by tissue transglutaminase (TTG) levels being over ten times the upper limit of normal, followed by similar repeat result and positive for HLA-DQ2/8 haplotype. Child therefore did not need a duodenal biopsy. First appointment since diagnosis.

History

- ○ Obtain history of symptoms, especially weight loss and fatigue
- ○ Ask about other autoimmune disease symptoms – particular associations are type 1 diabetes (5%), autoimmune thyroiditis, autoimmune liver disease
- ○ Ask about the impact this has already had on the child and family and if any other family members have the condition. Gluten-free products are expensive but a limited selection can be obtained on prescription
- ○ Ask about how much information the family has been given, or has sought for themselves.

Management planning – issues that may be discussed

- ○ Gluten-free diet for life. Should certainly avoid wheat, barley and rye, and most advised to avoid oats as can be contaminated in milling. This resolves symptoms, reverses bone demineralisation that may have occurred, decreases chance of pubertal delay. It also brings the risk (as a baseline, increased on gluten) of future gastrointestinal malignancy back towards that of the normal population
- ○ Dietetic input at diagnosis is very important – parents need regular advice, particularly around food labelling
- ○ Restaurants and supermarkets are now more attuned to needs of sufferers
- ○ Contact groups – encourage to join Coeliac UK (www.coeliac.co.uk)
- ○ Regular outpatient follow-up, looking at symptom resolution, growth and compliance to gluten-free diet – TTG checked (should return to normal after 3–6 months of treatment)
- ○ The diagnosis has been confirmed and is lifelong – this child would not be re-challenged or need biopsy providing symptoms and TTG return to normal on a gluten-free diet
- ○ Please refer to European Society of Paediatric Gastroenterology, Hepatology and Nutrition (ESPGHAN) guidelines, which clarify many issues, particularly around diagnosis. Note that many children still require duodenal biopsy particularly if TTG not ten times baseline, symptoms are non-specific or parents/clinicians make the decision that a biopsy is necessary.

3.2 Scenario two: insulin-dependent diabetes – morning hyperglycaemia in a 12-year-old

There are many patients with diabetes who are well known to paediatric outpatient departments and who are invited to the examination, in particular to this station. It is well worth attending specialist diabetes clinics to see the full range of problems with which children with diabetes and their families can present to the clinician. Be aware that insulin pumps are now more commonplace and can have their advantages, but also specific problems that can need 'troubleshooting'.

There should be detailed local guidelines covering diagnosis, ketoacidosis, 'sick-day' rules, hypoglycaemia, insulin pump difficulties and guidance about when a child should be assessed in hospital. NICE has produced guidelines on the diagnosis and management of diabetes which you should access and read.

History

- ○ When were they diagnosed?
- ○ Current treatment – including any prior manipulation to try to resolve morning hyperglycaemia
- ○ Compliance with treatment and indicators of control – clinic attendances, last HbA1c, frequency and pattern of hyperglycaemia, blood ketones often high? Diary of blood sugars and blood ketones kept regularly (look at diary for patterns if child has one)
- ○ Hospital admissions, previous hypoglycaemia (especially nocturnal) or diabetic ketoacidosis
- ○ Growth – is child thriving?
- ○ Impact on family and help they are receiving already
- ○ Co-morbidity – renal disease, coeliac disease, thyroid disease
- ○ Anything else the family feel is important.

Management planning – issues that may be discussed

- ○ Differential of this scenario:
  - Non-compliance
  - Dawn phenomenon (hyperglycaemia coinciding with early morning growth hormone secretion)
  - Inadequate insulin administration
  - 'Well-controlled' child experiencing *nocturnal hypoglycaemia*. Rebound hyperglycaemia occurs by the morning due to compensatory hormonal mechanisms. Such children may have had their bedtime insulin increased to theoretically bring down their morning blood sugars, with the opposite effect.

○ You have established that there is good control from your history. This child needs regular nocturnal blood sugar measurements (BMs) to confirm your suspicions of nocturnal hypoglycaemia. This problem is less common with basal bolus regimens.

○ Discussion of different insulin regimens and whether child should be considered for insulin pump therapy. If discuss pumps, be prepared to cover:
  - indications/eligibility
  - benefits
  - risks (eg pump failure with rapid ketosis if not spotted)
  - training for families and staff

○ Emergency management of hypoglycaemia

○ Emergency management of diabetic ketoacidosis (DKA)

○ Psychological issues around diabetes in children.

3.3 Scenario three: multiple 'seizures' in a teenager

You would need to take a focused history. The goal is to establish whether this is epilepsy, and if so what type of seizures have been occurring. If there have been a small number of events, try to describe each one from *before* the event to recovery. If there have been many similar-sounding events, try to elicit a description of a typical event. Remember that NICE has produced guidelines on epilepsy management (www.nice.org.uk/cg20).

The patient in this example has a primary generalised epilepsy. You may be given a patient with 'paroxysmal episodes', which are seizures that have a cause (eg febrile convulsions, reflex anoxic seizures). Remember that a child who has had 'seizures' does not always have epilepsy. Cardiac events such as arrhythmias are an important differential, as they can manifest as a collapse and a seizure secondary to hypoxia. It is important also to consider vasovagal episodes and 'Non-Epileptiform Attack Disorder' (previously known as 'pseudoseizures') in the differential.

History

Before the event

- ○ Witnesses; where and at what time? (may be the family have recorded an episode on a mobile phone)
- ○ Behaviour immediately before the event – what they were doing, whether they had been well, ask the child whether they felt, saw or smelled anything unusual
- ○ Triggers (eg flashing lights, pain, illness, noise, tiredness).

During event (some families are using mobile phones to record events, as mentioned above)

- ○ Full description of movements and noises. Were there automatisms (eg lip smacking)?
- ○ Eyes open? Any sign of awareness?
- ○ Unilateral features such as one-sided involvement/eye deviation?
- ○ Colour change, bladder and bowel incontinence
- ○ Total duration of event – often difficult for parents to recall
- ○ Did seizure terminate spontaneously or were anticonvulsants (± hospital admission) required?

After the event

- ○ Description of behaviour – levels of alertness. How long before child was back to normal?

General

- ❍ When did problem start?
- ❍ Are events getting more frequent? Associated morbidity, eg progressive headache or loss of skills, personality change
- ❍ Need to take a good birth, drug, developmental and family history
- ❍ Essential to elicit impact on schooling, family life, activities.

Management planning – issues that may be discussed

- ❍ The electroencephalogram (EEG) and its limitations (false-positives and false-negatives):
  - Different strategies for doing EEG – sleep-deprived, 24-hour …
- ❍ Always perform a baseline 12-lead ECG
- ❍ Consider MRI if concerns about space-occupying lesion, especially in this age group
- ❍ If a diagnosis of epilepsy is made, remember to consider the following and discuss with the child and family:
  - Lifestyle issues such as supervision, future impact on driving and DVLA restrictions (see Epilepsy Action website: www.epilepsy.org.uk/info/driving), contraceptive advice, sleep hygiene, career advice in older teenagers
  - Risks and benefits of anticonvulsant medication, and common side-effects of medications. See *British National Formulary for Children* (www.bnfc.org), the RCPCH *Medicines for Children* website and the aforementioned NICE guidelines
  - Referral to local epilepsy nurse specialist. Will need a care plan in place for home and school including when to give rescue medication (buccal midazolam) if has been deemed appropriate.

3.4 Scenario four: faltering growth in a 12-month-old

Weight has drifted from the 50th centile at 8 months to the 9th centile at 12 months.

History

- ○ Ask about the point at which weight became static; any preceding illness?
- ○ Food intake – has it changed, have there been problems with weaning? How much of diet still consists of milk?
- ○ Stool frequency (changed?) and consistency. Does stool ever contain blood?
- ○ Stool colour
- ○ Does infant vomit? If so, full description of character (ask about reflux symptoms), whether or not it is bilious, and frequency
- ○ Have there been any obvious allergic reactions to foods?
- ○ Are there chest symptoms or any other symptoms of chronic illness such as pallor, lethargy?
- ○ Any neurological concerns? Is development progressing normally?
- ○ Social history including support network for mother, how many other children to look after, possible maternal illness.

Management planning – issues that may be discussed

- ○ Differential diagnosis – such failure to gain weight can logically be split into the following categories:
  - Reduced intake – infant not being given enough calories (occasionally due to neglect)/refusing food
  - Malabsorption – such as that caused by pancreatic insufficiency of cystic fibrosis, or acquired enteropathy including post-infective, non-IgE food allergy or coeliac disease
  - Increased losses – diarrhoea and/or vomiting of acute or chronic illness, gastro-oesophageal reflux
  - Increasing energy expenditure – chronic illness, anaemia resulting from iron deficiency
  - Inefficient energy storage/release – metabolic disease
- ○ Be prepared to discuss non-organic causes of failure to thrive, as these are far commoner than organic pathologies
- ○ Investigations depend on the history elicited
- ○ Depending on symptoms and the most likely category based on the history and examination, first-line blood tests may include full blood count and film, ferritin, U&Es, thyroid function tests and tissue transglutaminase autoantibody screen for coeliac disease. Urine MCS should be sent. Consideration should be given to sweat testing even if the child has been

screened. A fasting glucose can be useful, and a metabolic screen should be considered

- Dieticians should be involved in all severe cases to assess the calorie intake, advise how best to increase intake and provide support for the family
- GP, health visitor and community nurse should also be involved
- Some patients and their families need an inpatient stay for further assessment and monitoring.

3.5 Scenario five: Down syndrome – yearly appointment, child now 11 years old and new to area

History

- Enquire as to child's general wellbeing and start to generate a 'problem list' – in terms of medical problems, many systems can be affected. What are the most significant problems for the child?
- In this case the child is hypothyroid and is on thyroxine, so ask specifically about symptoms of thyroxine over- or under-treatment. They have significant constipation, which is the child's biggest problem at the moment. Recurrent chest infections had been a problem previously, with one stay on ITU as an infant (pneumonia compounded by upper airway instability secondary to hypotonia).
- Ask about previous echocardiogram results; all children with Down syndrome should have been sent for an echocardiogram. In this case it was normal but remember the potential for future valvular dysfunction.
- Drug history, with attention to compliance and recent dose alterations.
- Was child under a paediatric endocrinologist in their previous area? How often were thyroid function tests (TFTs) being checked and when was last check?
- Immunisations up to date? (more vulnerable to infections, particularly respiratory) Remember winter influenza vaccine.
- Results of last hearing test? Both sensorineural and conductive hearing losses are much more common in Down syndrome; in fact around 50% of these children have hearing loss. The child may have a hearing aid.
- Visual problems? The child should have been checked for congenital cataracts, squint and refractive errors, all of which are more common.
- Ask about sleep, in particular snoring, agitation and apnoeas.
- Around one-third of children with Down syndrome have obstructive sleep apnoea. The child may have had polysomnographic sleep studies.
- Developmental history.
- Social history; in particular, ask about social and financial support. Any allowances?
- Schooling – currently in mainstream or special school?

Management planning – issues that may be discussed

It is useful to come up with a systems-based table/list of problems faced by children with Down syndrome. Evidence-based guidelines on the monitoring of many medical problems can be found on the website of the Down Syndrome Medical Interest Group (www.dsmig.org.uk). Some particularly important problems that are pertinent to this case are mentioned below:

- Plot child on adapted growth chart for patients with trisomy 21

- ❍ Management of hypothyroidism – usually 6-monthly thyroid function tests and initial communication with paediatric endocrinologist
- ❍ Remember that higher risk of coeliac disease (screen regularly), and type 1 diabetes
- ❍ Arrange an audiology review and ophthalmology review as necessary (should have been under surveillance in previous area) as high prevalence of problems such as congenital cataracts, squint, chronic otitis media with effusion, sensorineural deafness
- ❍ Discussion of higher risk of cervical spine disorders:
  - X-ray for atlanto-axial articular laxity if the child is symptomatic with neck pain, or has torticollis (holding the head in an unusual posture)
- ❍ Involvement of a wider team of health professionals based on need:
  - GP, community paediatrician, hospital specialists (ENT, respiratory, cardiology, orthopaedic, opthalmology, anaesthetics)
  - Physiotherapist and occupational therapist
  - Speech and language therapist
  - Audiologist, optometrist
  - Educational psychologist
  - Learning disability nurse

  Such professionals will meet regularly at a mutidisciplinary professionals meeting, often with a social worker.
- ❍ Who decides upon the level of educational support this child may need in secondary school?
- ❍ Parents should have been signposted to the Down Syndrome Association (www.downs-syndrome.org.uk)

3.6 Scenario six: severe constipation in a 6-year-old – first presentation to clinic

The majority (90%) of cases are functional (as is this case), but a focused history will need to be taken as other possible causes are Hirschsprung's disease, hypothyroidism, coeliac disease, spina bifida or meconium ileus equivalent. An exploration of the family's ideas, concerns and expectations (particularly around causes, tests, natural history of condition) is key in managing this condition. Many parents are aware of Hirschsprung's disease and you need to offer reassurance if there are no 'red flags' pointing to this condition.

History

- When did problem start (chronic if over 8 weeks) and were there any notable circumstances, eg illness, starting nursery/school, holiday to hot climate, difficult potty training?
- Ask about stool frequency and consistency; what do parents/child mean by constipation? Do they give a typical description of 'overflow' around impacted stool (+/– lack of awareness on passing overflow stool)? Are there withholding behaviours? Ask about night stools
- Is defecation painful +/– blood around stool (fissure)?
- Abdominal pain, often relieved on passing large stool?
- Vomiting? Persistent abdominal distension?
- Appetite, diet (ask about fibre intake) and fluid intake
- General health; enquire in particular about bladder problems, urinary tract infections, hypothyroid symptoms, behavioural problems. Is child thriving? Offer to look at growth chart in Child Health Record
- Any other health professionals, eg school nurse, involved?
- Medications; what has been tried? Sometimes the medication has worked well and has been stopped, only for the problem to recur
- Birth and past medical history – if term, was stool passed in first 48 h and were there problems as a baby (particularly first few weeks of life)?
- Family and social history impact of problem on family.

Management planning – issues that may be discussed

- Remember that such children need a full lower limb neurological examination, with a lower-back (ruling out obvious spinal abnormalities) and peri-anal inspection. Consider the possibility of peri-anal streptococcal infection
- Confident explanation of functional constipation, that have excluded red flags using history and examination (+/– full blood count, coeliac serology, thyroid function if pointers to perform these tests), and discussion of predictable difficulties such as overflow and soiling.

- A multidisciplinary approach works best
- Behavioural management – star charts, toileting
- Community management – school nurse, community children's nurse
- Consider referral to constipation clinics/nurse specialist if there is access to these services
- Dietary advice and stress the importance of fluid intake
- Regular attempts at toileting, advice on best posture
- Medical therapy – initial disimpaction with escalating doses of polyethylene glycol 3350 + electrolytes (eg Movicol) may be required. Admission is sometimes warranted for this if family finding it hard to administer. Try to avoid enema therapy as this can be distressing and can reinforce negative toileting experiences
- Try to settle child onto regular maintenance therapy, usually consisting of lower doses of Movicol. If there is a partial response to Movicol, addition of a stimulant laxative can help. However, if Movicol is not tolerated, substitute a stimulant laxative such as Senna together with a stool softener such as lactulose. The aim is to give the minimum dose that produces a regular, formed stool. It can take families a long time to get to this stage and they need to know that treatment can be required for many years. Medication should not be stopped abruptly unless there are significant compliance issues (would need review) or troublesome side-effects. In either scenario a review would be needed
- Refer to the NICE guidelines on the management of constipation in children and young people (www.nice.org.uk/cg99).

3.7 Scenario seven: multiple food allergies in a 5-year-old

History

- Child with egg, peanut and sesame allergy. One recent anaphylactic reaction.

Review appointment

- For each food reaction, take a history of:
  - Form in which food encountered and how much eaten
  - Whether the child was well at the time
  - Speed of reaction (most IgE-mediated food-allergic reactions occur within an hour, or at most 2 hours, after encountering the food responsible), and features of reaction, in sequence
  - In mild-moderate reactions most have urticaria, some angioedema presenting as facial swelling, some vomit +/– abdominal pain and loose stool
  - Always ask whether features of anaphylaxis:
    - Airway problems such as difficulty swallowing, stridor, very swollen tongue affecting airway
    - Breathing problems due to wheeze (+/– airway difficulties above)
    - Circulatory problems – hypotension (may have been measured in hospital) , collapse
  - Treatment administered and by whom (particularly if adrenaline was given)
- History of other allergic diseases – eczema, rhinitis, asthma. Some children will have all three to varying degrees. This patient has asthma
- Medication history; has an adrenaline autoinjector been prescribed? (two devices per medical kit recommended)
- Brief social history; can explore impact on child and family (do siblings eat offending foods?)
- Family history of food allergy?

Management planning – issues that may be discussed

- Testing – skin prick test (Spt) or specific IgE (spIgE), use 'cut-off' levels along with history to determine on follow-up whether child may be growing out of a food allergy
- Parental concerns – risk of serious reaction, how to avoid foods (especially when child in care of others), labelling (European Union law to label most common food allergens in ingredient list), parties and eating out
- Indications for adrenaline autoinjector – most obvious here being previous anaphylactic reaction to food, multiple food allergy and co-existent asthma (child on preventative inhaled steroid, still wheezes 3–4 times a week)
- Written management plans for home and school important

- ❍ Adrenaline autoinjector training very important
- ❍ Signpost to patient information – Anaphylaxis Campaign
- ❍ Medicalert bracelet.

3.8 Scenario eight: inflammatory bowel disease – management of relapse

Scenario of a 14-year-old boy with established Crohn's disease who has been referred to the rapid access clinic with bloody diarrhoea.

History

- ○ Age at diagnosis and how diagnosed
- ○ How many relapses so far?
- ○ Current symptoms pertaining to this relapse; include stool frequency, consistency, presence of blood and mucus, abdominal pain. Ask whether nocturnal symptoms in addition to daytime symptoms
- ○ Systemic symptoms including extra-intestinal manifestations – especially enquire about joints, eyes, skin
- ○ Evidence of weight loss? Check growth status (you may be shown a chart), including height velocity
- ○ Ask about pubertal status, as this is often delayed
- ○ Previous and current treatment (dietary, medical, surgical); ask about side-effects
- ○ Most recent levels of inflammatory markers, haemoglobin and albumin
- ○ Family history of IBD or other autoimmune conditions
- ○ Social history including effects of illness on family unit and schooling. Remember potential for considerable psychosocial morbidity.

Management planning – issues that may be discussed

- ○ Clinical assessment of unwell child with severe exacerbation
- ○ The use of markers of increased disease activity – raised erythrocyte sedimentation rate (ESR), C-reactive protein (CRP), white cell count (WCC), lowered albumin and haemoglobin, stool calprotectin
- ○ Home versus hospital management of relapses, and threshold for hospital admission
- ○ Discussion of main treatment approaches for relapses – exclusive enteral nutrition (polymeric/elemental feeds) or corticosteroids
- ○ You will need to know benefits versus risks of these treatments – remember that many patients become corticosteroid dependent or resistant
- ○ Involvement of a dietician is essential
- ○ Management of gastrointestinal complications of Crohn's disease, eg toxic megacolon (colonic dilatation > 6 cm), abscesses, fistulae
- ○ Indications for surgical treatment; remember that over 50% of patients will need an operation at some time in their life
- ○ Further treatments (particularly if steroid resistant and surgery not deemed suitable) to be aware of – azathioprine, 6-mercaptopurine, methotrexate. If

not responding to these, infliximab, adalimumab (monoclonal antibody) treatment can be considered – carries increased risk of malignancies.

In the UK, guidelines on the management of inflammatory bowel disease have been published by the British Society of Paediatric Gastroenterology, Hepatology and Nutrition (BSPGHAN) (www.bspghan.org.uk/documents/IBDGuidelines.pdf).

3.9 Scenario nine: headaches – 9-year-old child referred for further investigation and management

History

- For how long have headaches been going on?
- Patterns in relation to time of day/week
- Are there any factors that trigger onset?
- Description of onset, severity, duration and frequency
- Characteristics of pain – especially location and severity, and whether or not the headache is exacerbated by coughing, bending over
- Associated symptoms, eg vomiting, visual disturbance, vertigo or other neurological symptoms
- Ask the parents about the child's appearance and behaviour during an episode
- Are headaches getting worse or more frequent?
- Past medical history of note? Family history of headaches (especially migraine and variants)? There may be ideas/concerns/expectations around possibility of brain tumour, especially if family history
- Dietary history
- Social history, looking for any potential stressors
- Effect on schooling, family life.

Management planning – issues that may be discussed

- Discussion of differential diagnosis based on history (many patients with *migraine* would be suitable to attend this station; remember that migraine can be on a spectrum with tension headache)
- Tests in outpatient scenario – fundoscopy, visual fields, blood pressure, head circumference
- In the history, 'red flag' features of a headache that would warrant neuroimaging including the child being woken from sleep by the headache, morning/recumbent headache, occipital headache, headache that is worsened by coughing, bending over or straining at stool, increasing severity and frequency of headache, new onset of lethargy, or any change in behaviour and/or school performance
- Sinister clinical features – meningeal signs, altered conscious level, papilloedema (late sign), hypertension, lateral rectus palsy.

Management of migraine (child in this case has migraine clinically)

- ❍ Reassurance
- ❍ Discuss any exacerbating factors, eg poor sleep, tensions at home, bullying (this patient was being bullied which contributed to his migraine and tension headaches)
- ❍ Lifestyle and dietary advice
- ❍ Headache and food diary to try to capture exacerbating factors
- ❍ Pharmacological treatment:
  - Rescue, eg simple analgesia such as paracetomol or more powerful agents such as 5 HT agonist, eg sumatriptan, when headache first coming on
  - Prophylaxis, eg pizotifen, propranolol (side-effects such as tiredness, weight gain can be troublesome)
  - Anti-migraine diet – chocolate, cheese, caffeine, marmite free (or some similar regimen – talk with your dietician).

3.10 Scenario ten: eczema – management of flare-up in a 6-year-old

Eczema is common, causes much morbidity and can have an impact on families as well as the child. A good understanding of management and complications would be expected. Guidelines on eczema management can be found on the NICE website (www.nice.org.uk/cg57).

History

- When did child first develop eczema?
- Precipitants of flare-ups
- Severity and frequency of exacerbations, hospital admissions
- Current treatments – differentiate between creams and ointments, establish frequency of applications. Does child apply treatment at school?
- Enquire about courses of oral antibiotics
- Enquire about whether there is co-existent food allergy; child may have been on an exclusion diet. Did this work?
- Birth and past medical history; establish degree of atopy and any tendency for severe/recurrent infections
- Ask about home environment (house dust mite exacerbates problem), impact on sleep, schooling, impact on parents
- Family history of atopy very common (70%).

Management planning – issues that may be discussed

- Parental education
- Avoidance of soaps/wool/rough clothing
- Allergen avoidance (environmental/food allergies can exist but there are usually multiple factors involved, and allergen exclusion may not help as much as patient and family would like)
- Nail care
- Topical treatments:
  - Emollients for bath or shower; warn families that they can leave the bath very slippery
  - Soap substitutes, eg Epaderm® ointment mixed with water
  - Apply emollient directly onto skin, eg Doublebase® 3–4 times/day if possible
  - Topical corticosteroids – remember that regular intermittent treatment with potent corticosteroids is as safe and effective as long-term hydrocortisone use
  - Topical calcineurin inhibitors (eg tacrolimus) – especially useful on the face as there is no skin thinning. Remember side-effect of burning sensation. Need sun protection if on sites exposed to sun

- Referral to paediatric dermatologist for difficult-to-treat eczema, eg if considering topical calcineurin inhibitors.

Other issues

- Bandaging – wet and dry wraps
- Treat any suspected bacterial infections topically or with oral antibiotics such as flucloxacillin for moderate to severe infected eczema. Take skin swabs first and give a 10- to 14-day course
- Recognition and treatment of eczema herpeticum: families should be shown images of secondarily infected eczema to aid recognition of the condition, so that they can seek help urgently if it starts to develop
- Indications for hospital admission. Remember that some families can cope with severe flare-ups better than others and that a written action plan/telephone advice can be useful.

3.11 Scenario eleven: chronic fatigue syndrome (CFS) – 14-year-old girl referred to clinic

Commonly seen in outpatient departments. NICE has published guidelines on CFS in adults and children on its website (www.nice.org.uk/cg53).

Remember that CFS/myalgic encephalomyelitis (ME) in children is defined as generalised fatigue that persists for more than 3 months, after routine tests and investigations have failed to identify an obvious underlying cause. There are currently no specific diagnostic criteria.

History

○ Gradual/sudden onset of problems. Often a preceding illness (Epstein–Barr virus, influenza, pharyngitis)
○ Explore all symptoms – cardinal feature is *debilitating physical and mental fatigue, worsened by physical and mental activity*. Headaches, sleep disturbance, difficulty concentrating, myalgia and arthralgia can occur
○ General health and past medical history, medications
○ Psychiatric history; ask about current or previous depression/anxiety
○ Impact on child and family, especially school days missed, family activities compromised
○ What measures have already been taken to aid in recovery?

Management planning – issues that may be discussed

○ Importance of thorough physical examination should be mentioned
○ Differential diagnoses – anaemia, autoimmune disease, chronic infection, endocrine disorder (hypothyroidism and diabetes), depression, drugs, sleep disorder such as narcolepsy
○ Once a positive diagnosis has been made, agree a management plan with the family and other relevant health professionals, such as the child's GP and OT/physiotherapist. The patient may require specialist referral
○ Stress that family need a co-ordinator of care
○ Inform Local Education Authority (with family's consent) where applicable
○ Methods of establishing baseline activities – patient should gradually increase activity, with regular reviews. Warn patient that there will be 'payback' symptoms a day after excessive activity
○ Recording baseline activities, more strenuous or stressful activities and flare-ups ('payback' symptoms) with diary.
○ Advice on diet/sleep, referral for co-morbidities such as psychiatric symptoms. Some patients benefit from cognitive-behavioural therapy (CBT)
○ Goal is gradual reintroduction to school and return to normal activities.

3.12 Scenario twelve: review of patient with extrahepatic biliary atresia post liver transplant

This is the kind of patient who would possibly be used in the clinical abdominal station on the same examination day and whose clinical features are covered in Chapter 9.

History

- Age and mode of presentation – nearly all will have had prolonged jaundice, pale stools, dark urine
- Associated malformations – splenic, cardiac, gut (malrotation) most commonly
- Investigations to confirm diagnosis: liver ultrasound ± hepatobiliary iminodiacetic acid (HIDA) scan (to evaluate whether there is obstruction to normal flow of bile), liver biopsy
- Early complications, eg coagulopathy
- Which specialist centre has managed/manages the patient?
- Age at initial Kasai portoenterostomy? Better results before 8 weeks in several case series
- Complications of surgery, eg ascending cholangitis
- Reason for liver transplant; ask if child had oesophageal varices and/or liver failure
- Discuss post-operative period and complications
- Drug treatment post-transplant – especially immunosuppression and complications
- Immunisations
- Ask about intervals of follow-up, involvement of other health professionals, eg dietician, specialist nurse
- Social history; acknowledge impact on family (such children spend long periods in hospitals that are often far from their home).

Management planning – issues that may be discussed

- Outpatient management of prolonged jaundice – when to investigate, what to rule out
- Discussion of Kasai procedure and indications for later liver transplant
- Management of ascending cholangitis and liver failure
- Working a child up for a liver transplant – main considerations and awareness that donor supply is one of the main limiting factors
- Immunosuppression, eg with tacrolimus, and general management including immunisations.

PART II

The communication skills station

4

INTRODUCTION AND SUGGESTED APPROACH

This section of the exam is intended to look at how you communicate with patients and/or their parents in your everyday clinical practice. You are awarded marks for both your communication skills and clinical knowledge, and so it is not enough (although is essential) to just be a good listener – your job is to address patient's and their family's concerns appropriately and to provide them with reliable, accurate information about the child's condition and the ongoing management plan. This requires a broad knowledge of common paediatric conditions similar to that required in the written part of the MRCPCH exam. In preparing for this station you should concentrate as much on your background knowledge as you do on your communication style.

It is worth reading through the RCPCH Anchor Statements as well as the mark sheets (on the RCPCH website) as you prepare for this section of the exam.

You are being marked in three areas:

1. Conduct of interview
2. Appropriate explanation and negotiation
3. Accuracy of information given.

The six main patterns of communication scenario described by the College are:

1. Information giving
2. Breaking bad news
3. Consent
4. Critical incident

5. Ethics
6. Education.

Here are some suggestions for how to prepare for this section:

1. Clear guidance about expectations and some comprehensive examples of cases are available from the RCPCH (www.rcpch.ac.uk) and reviewing this information should be your first step.
2. Make a shortlist of conditions that you plan to know in detail (see Chapter 6 'Further Scenarios'). You should know about the usual treatments offered and practise how you would talk through these clearly and without using any medical jargon. No conditions should be considered too serious or too trivial to come up in the exam.
3. A good way of practising suggested approaches would be to ask a colleagues to watch you explain management decisions to the parents during a ward round. Give them a copy of the Anchor Statements for the communication skills section. Ask for feedback and suggestions for improvement. Involve parents in the feedback process if you feel comfortable doing this.
4. Role play with colleagues who are also preparing for the exam. Prepare scenarios for each other. Make sure you practise the scenarios you feel least comfortable with. Practise under 'formal' conditions and practise keeping to time.

SUGGESTED APPROACH

All of the scenarios will fit into one of the following broad categories. This is not intended to be a syllabus for studying for the communication skills section, but a framework (after initial introductions) to structure your approach to these stations. This should make you feel much more confident in the run-up to the examination.

Information giving

Talking about chronic conditions

Frequently this takes the form of talking to a child or parent about a change in management because of a clinical deterioration. Parents and or children may disagree with the medical team's interpretation or plan. When parents or patients disagree with the team or need more convincing, a suggested approach is to:

- Ask them what their concerns are

- ○ Discuss each concern: refer to any evidence you know that backs up a sensible medical plan (which could be published articles or national guidelines) or describe what is generally accepted practice
- ○ Talk through the risk/benefit of any alternative management strategies
- ○ When there is potential for harm to the child if there is no agreement, then describe plainly what harm could arise, how likely this is to happen and the expected time frame
- ○ If they remain unconvinced, offer to take their concerns back to the team looking after them and arrange another time to discuss the issue further
- ○ Summarise what you have talked about, including concerns that have been raised and what was decided.

Explaining a new treatment

- ○ Check patients' and parents' understanding of current status and need for change in management
- ○ Describe what the new treatment is
- ○ Explain how it works (use diagrams wherever possible)
- ○ Explain how it is administered
- ○ Explain what the desired/expected outcome is
- ○ Explain how long the patient will need to take it
- ○ Talk about side-effects, how likely they are, how to recognise them and what to do about them
- ○ Give an opportunity for questions and explore any concerns that the patient or their parents have about the new treatment
- ○ Summarise what you've said and check parents' understanding of this:
    - Discuss how the family can access/give written information where possible.

Breaking bad news

- ○ After initial introductions swiftly move on to talking about new information, ie give the diagnosis
- ○ Establish their prior knowledge of this diagnosis – do they know anyone with this condition
- ○ Talk about the implications of the new information
- ○ Allow space for patient/parent to respond, express feelings, ask questions
- ○ Answer concerns honestly and sincerely
- ○ Talk about what is going to happen now
- ○ Summarise, giving the opportunity for more questions
- ○ Offer to meet again to go through any questions
- ○ Offer written literature, for example contact details for support group if appropriate.

Consent

Seeking consent

- ○ Check parents' understanding of their child's current health/condition. Acknowledge any concerns/anxiety that they are experiencing
- ○ Explain the need for the procedure
- ○ Explain how it is done
- ○ Explain the anticipated outcome
- ○ Talk through any risks (be realistic about what parents should be told) and what will be done to minimise these risks
- ○ Talk through any alternatives
- ○ Give an opportunity for questions and address these immediately
- ○ Summarise what you've said and what has been decided, and check parents' understanding of this.

> The Anchor Statements specify that you will need to demonstrate that 'steps have been taken to ensure that important messages have been understood'.

Critical incident

Telling parents about a medical error:

- ○ Apologise in a sincere manner. Most angry parents will become angrier if they sense they are not going to get an apology or that the apology is insincere. Get a colleague to critique your 'apology' style.
- ○ Be prepared to discuss the following with the parent:
  - what went wrong
  - what the likely risks are to the patient
  - what harm has already been done
  - what you are going to do about it to keep the patient safe now
  - what has been done so far about the mistake
  - what you will do to make sure this won't happen again.
- ○ Be aware of your hospital's complaints process and critical/serious incident monitoring procedures.

Duty of candour

Included within this topic is the Duty of Candour. Candour can be defined as: 'The volunteering of all relevant information to persons who have or may have been harmed by the provision of services, whether or not the information has been requested and whether or not a complaint or a report about that provision has been made.'

The new statutory duty of candour was introduced for NHS bodies in England (trusts, foundation trusts and special health authorities) from November 2014, and applied to all other care providers registered with the Care Quality Commission from 1 April 2015.

Candour is about being completely open, honest and truthful in all our dealings with patients and to provide full answers to any questions related to treatment. The threshold for discussing a situation with a patient can be a grey area (as in the case later in this chapter). One way of looking at it would be to turn it around and ask yourself if you were the patient would you want to know these details? In most cases the answer is yes; patients have entrusted us with their health and well-being, and we need to honour and respect that.

Ethics

Having an ethical discussion about a case where the correct course of action is unclear.

Prepare a general ethical framework that you can apply to many different situations. A suggested approach is discussed in detail in the cases in Chapters 5 and 6.

Telephone conversation

Your communication skills task may involve conveying information on the telephone to a parent, doctor or other professional. In the room you will be sitting on one side of a screen, talking to someone on the other side, with the examiner sitting at one end of the screen so that they can see both of you. The tasks given will reflect the kind of telephone conversation that a paediatric registrar would be expected to handle during everyday clinical practice. They will still fall into the six main patterns of communication described above. Examples which are appropriate to a telephone conversation station include:

- Calling a parent about their child's investigation results (information giving)
- Asking another professional (eg a GP or health visitor) to participate in a child's follow-up or ongoing care (information giving)

- Talking to a parent who has phoned to make a complaint (critical incident)
- Talking to a GP who has phoned to ask for advice about a patient (information giving).

Key differences between a telephone scenario and other communication skills tasks:

- Your opening remarks set the tone. You will pick up points for a 'full greeting and introduction'. Always introduce yourself by clearly stating who you are, where you are calling from and what your role is. You should then clarify who you are speaking to. For example, 'Hello, my name is Dr Smith, I'm one of the paediatric registrars calling from City Hospital. I would like to talk to Claire Anderson's mother. Is she in?'
- Whenever you are phoning someone to talk about something important you will need to check whether they are able to listen at that time. Don't forget that normal telephone etiquette applies in the exam! The most important first question to put to a patient or parent is therefore, 'Is now a good time to talk?' This alerts them to the fact that you have something important to say which may take more than a few seconds. If you are phoning a doctor or other professional, you could ask 'Have you got time to discuss this now?'
- You will not receive any 'non-verbal cues', so you will need to show that you are actively listening in other ways.
- In clinical practice, all patient-related telephone calls should be documented in the patient's clinical notes. When you finish the call, you should aim to let the patient or parent know that you will be documenting the conversation. You could offer to send them a letter confirming what you have spoken about, or tell them that you will write it down in the notes so that their consultant knows what was discussed when they are next seen.

Education

A significant part of our job as doctors is to educate – from teaching our patients about their diagnoses and how to manage them, to teaching medical students or junior colleagues about how to carry out an audit.

It is worth thinking about the different ways you might approach this task depending on who you are delivering the education to. The framework is similar to the other categories that fall into the communication skills scenario and starts with:

- Check the person's understanding of the topic
- Give a broad overview of the topic, then focus in more detail on any points that are particularly relevant

- Give an opportunity for questions
- Summarise what you've said, and check understanding.

There are examples of possible scenarios in chapters 5 and 6.

Suggestions for how to prepare yourself during the exam

- This is a 9-min station consisting of spoken interaction. You will have up to 2 minutes before entering the room and starting the assessment to read about the task and prepare yourself.
- Use the full 2 minutes to read the scenario. Make notes on key points that you need to get across as well as making notes of any names given – try and use the names of the patients/parents during the scenario. You may want to use your pencil and paper to draw diagrams if explaining a diagnosis – such as vesico-ureteric reflux.
- Try to put yourself in role: imagine yourself in this scenario in the right setting – your natural communication abilities are more likely to come across if you picture yourself in a real environment.
- Remember you have been given the relevant information you need – you are not taking a history.
- Once you have introduced yourself, ('Good morning my name is Dr Chris Smith; I am one of the neonatal registars'), it may be appropriate to set an 'agreed agenda' with the parent, to structure the consultation.
- Pause to listen – remember you tend to talk faster when you are nervous.
- You will get a warning when there are 2 minutes to go. You need to think about summarising what has been talked about, asking if they have any further queries and bringing the 9 minutes to a satisfactory conclusion.

5

CASES

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5.1 Case one: breaking bad news – new diagnosis of Down syndrome

The following is a fully worked-through example of a scenario that is typical of the sorts of cases you will be expected to deal with in the communication skills section. In keeping with the layout that you will encounter in the exam we have included boxed 'instructions to the candidate' as well as those given to the role player and examiner (who has a copy of all three 'instructions'). These are similar to the instructions seen within the specimen scenarios on the RCPCH website, www.rcpch.ac.uk. The intention is that this, and the following scenarios can be used as templates so that you can develop a structured approach to this section of the exam, and become familiar with the level of background knowledge you will need to be fully prepared. This scenario could fit into the category of 'Breaking bad news' but will also test your ability to give information in layman's language and requires a working knowledge of Down syndrome.

Instructions to candidate

○ This station is a test of your ability to convey information based on the specific task given.

> ***Timing:***
>
> *Total time provided for this station is **9 min**.*
>
> *There are **2 min** before you are called in to read this sheet and prepare yourself. A bell will signal the start of the station and you will be invited into the examination room – take your instruction sheet with you.*
>
> *You will have **9 min** in total for the consultation; a warning will be issued when you have **2 min** left.*
>
> *You will not be assessed on the speed at which you communicate; you may find that you run out of time to finish the consultation.*
>
> ***You may take notes.***
>
> ***You will not be asked to examine the patient.***

Role

A Specialist Trainee in paediatrics, working in a district general hospital.

Setting

Side room on the postnatal ward.

You are talking to

Mother of Baby Williams, a baby boy born overnight; he is currently almost 10 hours old. You have just examined him in one of the side rooms on the postnatal ward, as your SHO is concerned he may have Down syndrome. You feel that clinically, based on your examination, he does have Down syndrome.

Other information

Baby Williams was born at term following an uneventful pregnancy, his mother is 27 years old and based on the 'triple test' and nuchal thickness USS, she was not at ncreased risk of having a baby with Down syndrome. This is her and her husband's first baby.

Task

- To explain the likely diagnosis
- To explain further investigations required
- To explain the implications of the diagnosis (ie what is Down syndrome and how it affects children)

You are not expected to gather any further medical history during this consultation.

Role player information

This is the information that is provided to whoever is playing the role of the parent.)

Total time provided for this station is 9 min.

There will be a dialogue between you and the candidate only; you will not communicate with the examiner during the 9-min period.

Role

ou are Jenny Williams and have just given birth to your baby Benji who in your eyes perfect. Up until this point you have had no concerns about Benji. This is your first aby. You have been waiting a long time for this baby – and have had two previous miscarriages.

ou have heard of Down syndrome. You don't know anyone who has it, but have een people on television with it, and don't really have any idea about it apart from he fact that Down's children often have 'special needs' at school.

Your general feelings

You are feeling more and more anxious – you can tell something is very wrong. You want to know what it is and how to make it better. The midwife asked you to call your husband in, but you do not want to wait for him to get in to the hospital and just want the doctor to tell you straight away what is going on.

What to expect from the candidate and what your feelings and *possible* further questions could be

○ The candidate should explain to you that they think Benji has Down syndrome.
○ The candidate should explain to you how they will confirm the diagnosis (ie by doing a blood test)
○ You should express that you do not know anything about Down syndrome
○ The candidate should give you a basic explanation of Down syndrome – how it will effect Benji's development, and other associated problems that will need to be screened/tested for.

After the candidate has explained what Down syndrome is your further questions might include

○ How will this affect his future (schooling, keeping up with other children)?
○ Will he ever get better from this?
○ How could this happen when all your screening tests in pregnancy were normal?
○ You and your partner are hoping to have other children; will they be affected too? How did he get this condition; did you give it to him?

Examiner information

Total time provided for this station is 9 min.

There will be a dialogue between the candidate and the role player only. Please warn the candidate when only 2 min of the consultation remain. Otherwise do not communicate with candidate or role player during the 9 min consultation.

If the candidate finishes early, you should check that they have finished. If so, they should remain in the room until the session has ended.

Guide notes towards Expected Standard

○ Explain that based on your examination Benji has many of the features of a baby with Down syndrome
○ You will need to do a blood test to confirm the diagnosis

○ Appropriate summary of Down syndrome, including its association with congenital heart defects that will need to be screened for.

Suggested plan of attack

Preparation

In this scenario you are expected to give relevant information in an effective way, but you are also breaking bad news. A breaking bad news 'checklist' which could be adapted for this situation is given in the Box. Remember that in real clinical practice when you are about to discuss something with a parent that you suspect will be distressing or sad for them you need to think about the following:

○ Environment: private and distraction free
○ Interruption free: hand over your bleep, tell your team where you are and what you're doing
○ Are the right people there? This includes relevant medical staff (in this case the midwife looking after Benji and his mother) and the parents'/patient's support network. Are you the right person to be talking about this now (should your senior colleagues be involved)? A senior colleague should certainly be aware that you suspect Down syndrome and are having this discussion
○ Do you know all the facts: not only about the patient's background and medical history but about the condition you are talking about and how it is managed in your hospital.

Breaking bad news checklist

○ Establish common knowledge base
○ Talk about new information
○ Talk about the implications of the new information
○ Allow space for patient/parent to respond: expressing feelings, asking questions
○ Answer concerns honestly and sincerely
○ Talk about what is going to happen now
○ Summarise with opportunity for more questions
○ Offer another meeting
○ Offer written literature, contact details for support group if appropriate

Note how the 'task' instructions appear brief and relatively straightforward; however there are clearly several components to this task and all of them are being specifically watched for by the examiner. In the preparation time you should make a quick list of what you feel needs to be said to fully satisfy your task of 'explaining implications'. This could look like this:

1. Establish how much mother already knows (in this case about Down syndrome).
2. Confirmation of diagnosis, describe what the condition is and (very broadly) the clinical features.
3. Discuss associated conditions to be screened for (eg echo for congenital heart disease, hypothyroidism), potential problems/complications.
4. Support groups/useful websites.

Introduction

You might like to start by saying to the examiner that you would have already handed over your bleep and arranged for a midwife to be present with you (before you say anything to the role player).

When you introduce yourself, say where you fit into Benji's care. Establish a common knowledge base.

> *'Good morning Mrs Williams, my name is Dr Simpson. I'm the specialist trainee in the team looking after Benji. Having just examined Benji I feel he has some features which make me think he may have Down syndrome. Have you heard of Down syndrome or know anyone with it?'*

Explanation

Try to give the diagnosis as soon as possible with limited pre-amble. After you have said it, allow some time for it to sink in. Don't be afraid of silence – what seems like a very long awkward pause to you may not seem like a long time to the parent. You will need to find a way of allowing the parent to set the pace of the discussion following this pause so that you can explain the key points on your list of information to be conveyed.

Closing remarks

You need to check that she has understood one or two key points from the consultation and establish a plan for what will happen now.

> *'Mrs Simpson, I realise that I've given you a lot of information and that it is a lot to take in all at once. The main thing to remember is that I think Benji has Down syndrome. This is something that will affect his development as he grows up, though it is difficult to predict to what extent, it is also linked or associated with some other problems that we need to check Benji for.'*
>
> *'What is going to happen now is that I am going to do Benji's blood tests to confirm the diagnosis. I should have this result within 2 working days. In the meantime I will arrange for Benji to have an echocardiogram which is an ultrasound of his heart. Before going home we should have the results of these and*

> *also make sure Benji is feeding well. I'll also give you some patient information about Down syndrome including contact details for a website where you'll be able to find some useful information, Members of the team will be available to answer any questions you will have. We will let your GP and health visitor know.'*

A useful website to look at both for yourself and to pass on to parents is www.downs-syndrome.org.uk.

5.2 Case two: discussing an ethical dilemma – disclosure of HIV status

Candidate information

Background

You are a Specialist Trainee in paediatrics. You have just finished seeing a patient on the ward round who was admitted several days ago with tuberculosis. She is 11 years old and has recently arrived from Africa with her parents. She speaks no English but her mother speaks English well. Her mother is HIV positive but has not told her family. The patient was tested and her test has also come back positive. When her mother was told, she said that she did not wish her daughter to know. A medical student who was observing the round now wants to talk to you about this patient.

Task

Discuss with the medical student the ethics behind the decision not to tell this patient that she has HIV.

Role player information

You are a final year medical student. You strongly believe that children should be fully involved in all aspects of their care and cannot understand why this information is being withheld.

Questions you should ask

- ○ Don't we legally have to tell her about it?
- ○ If she is not to be told now, when is she going to be told?

Questions you may ask

- ○ What about her father? Does he have a right to know? Could he decide to tell her?

Examiner information

Guide notes towards Expected Standard

- ○ The candidate is aware of reasons for and against informing the child of her HIV status

- The candidate is able to explain these clearly and confidently
- The candidate shows respect for others' views.

Suggested plan of attack

Preparation

This is a scenario where you are expected to explore the ethics behind a difficult situation. As with all these sorts of scenarios the examiner will be looking to see that you are able to understand both sides of an argument and that you can identify the issues involved, particularly those concerning conflicts in interests or rights. You will need to demonstrate your ability to do this and you have only got 9 min! It is unlikely that there will be any right or wrong answers, although if there are any legal implications or RCPCH/General Medical Council guidance about an issue, it is essential that this enters into your discussion. A list of key documents which have influenced this area is provided in a Box at the end of the notes to this case.

Suggested structure for an ethical discussion

- Be clear about the question being asked.
- List 'arguments for' and 'arguments against'. As a way of categorising the various conflicting interests you could use the traditional medical ethical framework (autonomy, beneficence, non-maleficience, justice), but make sure you include some explanation of this as you will probably be talking to a medical student or junior colleague who may not have experience of discussing things in this way. The concepts behind the ethical framework are summarised in the Box in the notes at the end of this case.
- Decide on a way forward – this could be a practical way of resolving some of the conflicts identified. Examples could include agreeing another opportunity to discuss things further, involvement of senior colleagues or the hospital ethics committee, seeking advice from specialist groups or looking for published evidence on the topic

Using the suggested structure given in the Box, your list for the scenario could look something like this:

1. Main question, 'should an 11-year-old be told that they have HIV?'
2. Arguments for and against:
   - For:
     - 'Autonomy' – right to be actively involved in her own treatment

        - 'Doing good' – able to offer more support if diagnosis known, can help to develop coping strategies
        - 'Do no harm' – protecting her future sexual partners
    - Against:
        - 'Do no harm' – less control over who knows, potentially exposing her and her family to stigma if she tells someone else, Mum's right to confidentiality
3. Way forward:
    - Explore reasons why Mum does not want her to know
    - Negotiate phased disclosure
    - Offer support.

Introduction

You need to establish clearly what you are discussing and to seek out the medical student's views.

> *'That was a really difficult case. Deciding whether or not a child should know that they have HIV is a difficult decision to make. What do you think is the right thing for this mother to do?'*

Explanation

You need to introduce a structured way of weighing up all the options (point 2 above, 'Arguments for and against').

> *'What do you think are the reasons in favour of her knowing that she has HIV?'*
>
> *'Can you think of any disadvantages for her or her mother if she is told that she has HIV?'*
>
> *'One way that I have found helpful in weighing up difficult decisions is to think in terms of a doctor's obligations; for example, autonomy, doing good, doing no harm and treating people equally.'*

Reasons for telling

Autonomy:

- The patient has the right to access support and information to help her understand her illness
- She has the right to be actively involved in her care.

Beneficence:

- Reduce anxiety
- Compliance with medication will be important – she will want to know why she has to take medication/come to clinics and have blood tests
- She may become sexually active in near future and will need to protect future partners.

Reasons against telling
The mother knows her own child; the balance of benefits versus drawbacks cannot be made without reference to her views, including cultural issues on what her child needs.

Non-maleficience:

- The mother has a right to confidentiality
- Knowledge of her HIV status may alter her self-esteem
- Knowing that she is HIV positive may not improve her sense of wellbeing
- Protecting her from stigma at school.

Closing remarks

Try to find a practical way forward or 'next step' (point 3 above).

> *'It seems as though this mother will have major difficulties to face when she does decide to tell her daughter about HIV. It may help if when we next talk to her we outline the benefits to her daughter of knowing her diagnosis. We could also explore the reasons why she doesn't want her to know. There are lots of places where she could get some more specific advice and support. This hospital has some HIV specialist nurses who are really experienced in talking through these issues with parents. There are also lots of support groups and websites where she can get some really practical help and advice for how to go about telling her daughter.'*

Background notes

Ethical framework

This is an aid to decision-making when faced with an issue where the 'right' course of action is unclear. Four different domains offer different ways of looking at a doctor's duties or obligations. When a difficult issue is defined in terms of these duties the fundamental conflicts that are present become more explicit. This helps the clinician who is weighing up different options to view the issue from various different perspectives and to come to a clear decision/recommendation/stance that they can then justify.

The four domains are described in the Box. Not all four domains will be relevant to each case you meet. However, being able to define a difficult problem in terms of these specific categories can help clarify the decision-making process.

The four domains of the traditional medical ethical framework

1. Autonomy – 'Deliberate self-rule'

One of the things we like as individuals is to be able to think for ourselves and make our own choices. Ideally these choices should be based on logical and rational thought, having been presented with all the information required. Things that detract from autonomy include:

- ○ Paternalism
- ○ Lack of capacity (eg mental illness)
- ○ Pressure from society (eg peer pressure)
- ○ Lack of self-confidence

As a medical professional we can increase a patient's autonomy by:

- ○ Asking for and valuing their opinion
- ○ Seeking to remove external pressures
- ○ Giving timely and clearly understandable information
- ○ Enabling good communication with health professionals

An individual's right to decide for themselves is ultimately limited by:

- ○ The law
- ○ Others' rights and safety
- ○ Limited resources

2. Beneficence – 'Doing good'

This is the obligation to do whatever will deliver the greatest benefit to the patient in terms of their present and future wellbeing. It also necessitates balancing up the risks and benefits of the different options.

3. Non-maleficience – 'Do no harm'

A doctor has an obligation to avoid causing harm. Harm is something that undermines a patient's wellbeing or limits their potential. Harm from medical treatment may occur as a recognised risk of a particular course of action or may be accidental. Harm can be physical, mental, emotional or spiritual. There are some harms so unacceptable to an individual patient that they will take whatever course of action necessary to limit the risk of their occurring.

4. Justice – 'Treating people equally'

This looks at the obligation to divide up resources fairly. How to judge what is fair is fraught with problems. There are advantages and disadvantages to distributing resources on the basis of societal values, evidence base, need or health economic measures such as QALYs (Quality Adjusted Life Years).

Telling a young person they have HIV

Once a child has been diagnosed with HIV most professionals would recommend informing them in a staged approach, starting with simple explanations and building up knowledge gradually. How and when they are told will depend on their age and development, their previous experience of HIV (for example in family members) and the need for treatment. In general the 'HIV' label is shared with children when they are about 12, but most will have prior knowledge of the basics of their condition, ie that it involves cells in their blood which are an important part of their immune system. It is essential they know their diagnosis and its implications before becoming sexually active. Learning about living with HIV is an ongoing process and information will need to be revisited and adapted as a child's needs and circumstances change.

There are many reasons why a parent might be apprehensive about their child knowing that they have HIV. A parent might have fears about who else their child will tell. They may worry how their child will cope with the information and whether it will affect their relationship. They may have feelings of guilt or fear, or may be struggling to deal with their own diagnosis. There are many support groups and websites offering excellent resources for parents who want to talk to their children about HIV (eg Children's HIV Association, www.chiva.org.uk). If the parent feels they will become too upset talking about it, they could arrange to be present when a trusted health professional talks to their child. In general the benefits of a parent communicating openly with their child outweigh the upset or other difficulties that it may cause. The child may already be exposed to misinformation about HIV through the media and their peers. They may be on a lot of medication with significant risks associated with non-compliance. They will need time to develop coping strategies, and they may have a degree of anxiety and may feel more in control if they have more information. There are picture books available that parents and medical staff can go through with children to aid their understanding of this condition. They explain why the child has to go for blood tests and why they need to keep taking their medications.

HIV is not a notifiable disease and there is no legal need for the school to know. There is no evidence that there has ever been any HIV transmission in school. In general it is recommended that only their head teacher and one other teacher is informed, so that they can have some pastoral support if needed.

Confidentiality

Much of law about disclosure of health information concerns sharing information when a child is at risk of or has suffered abuse. In these cases the child's welfare is paramount and any threat to this needs to be taken seriously. Guidance is available from the General Medical Council and the Royal College of Paediatrics and Child Health:

- Confidentiality must not stand in the way of protecting the child
- Consent is normally required to disclose confidential information – this applies to information given by children in confidence
- Information from a child can be disclosed without the child's consent if the clinician believes it is in the child's best interests (for example, to prevent them suffering further abuse), but the clinician will need to be able to justify this
- It is good medical practice to inform the patient if you are going to pass on any information; tell them who you are passing it on to and why
- If you decide disclosure is not in the best interests of the patient, you should talk to a senior colleague and be prepared to justify your position
- Documentation is vitally important.

Key documents

The following is a list of helpful websites and key documents that influence the way we interpret ethical dilemmas in paediatrics. You need to be broadly familiar with these as they all influence the way paediatrics is practised in the UK:

- UK clinical ethics network (www.ukcen.net) provides up to date reliable information, policies and guidance on ethical issues
- The General Medical Council website has a number of helpful documents including the following (www.gmc-uk.org):
  - *Seeking Patient's Consent: The Ethical Considerations*
  - *0–18 Years: Guidance for all Doctors*
- RCPCH publications (www.rcpch.ac.uk):
  - *Making Decisions to limit treatment in life-limiting and life-threatening conditions in children: a framework for practice* (adc.bmj.com/content/100/Suppl_2s1.full)
  - The United Nations Convention on the Rights of the Child (www.unicef.org/crc)

5.3 Case three: an education task – explaining how to carry out a quality improvement project

Candidate information

Background

You are a Specialist Trainee in paediatrics working in a district general hospital. A foundation trainee comes to you asking for advice. He has been asked to perform a quality improvement project but not been given further guidance about how to go about this. He has heard of quality improvment but has not been involved in a project before.

Task

Explain the quality improvement/'PDSA' (Plan-Do-Study-Act) cycle to the foundation trainee.

Role player information

You are a foundation trainee. Your involvement in previous quality improvement (QI) projects has been limited; you remember having lectures at medical school about the subject and have seen a few presentations since qualifying. You now have to think of, and carry out a QI project and do not know where to start.

Questions you should ask

○ Why do I need to do this?
○ What project should I do?

Questions you may ask

○ Who can help me with this?
○ Will doing this project change anything?

Examiner information

Guide notes towards Expected Standard

○ Establishes level of prior knowledge
○ Clear and confident explanation
○ Ability to explain what a QI project is and why we do them
○ Checks understanding at the end of the discussion.

Suggested plan of attack

Preparation

This is an educational task and is checking not just your knowledge of the topic but your ability to teach it to another doctor. You should practise a clear structure for these sorts of tasks, which you can then apply to whatever subject is required.

A suggested approach to an educational task

In your preparation time, write down these headings and list points you need to get across:

1. Establish common ground. Find out what they already know
2. Agree a learning agenda. You could use phrases like, 'Would it be helpful if I explain to you ...?'
3. Outline the importance of what you are going to explain. Try to enthuse the person you are teaching, make it relevant to them
4. Explain it concisely and simply (you haven't got much time!). Use diagrams wherever possible
5. Summarise what you've said and check understanding. You could use phrases such as, 'To summarise, tell me: what are the main points to remember ...?'

Introduction

Aim to find out how much experience the trainee has had with quality improvement projects. You can then base your explanation on knowledge they already have. You need open-ended questions. Use the first few minutes to clearly establish what you are going to talk about and why it is important.

> *'I understand that you need to do a QI project. What do you know about QI projects? Would it be helpful if we spent some time talking through the process?'*

Explanation

Try not to make this a lecture but a conversation. Keep checking that what you are saying makes sense to the listener. Try to give a structure by using a diagram, or a list of key points, and make this structure explicit.

> *'The difference between audit and quality improvement is ... It is often easy to identify problems within the organisation or department in which we are working. However this is an opportunity to improve things.'*

'It might be worth thinking firstly "what is quality?"

- *safety*
- *effectiveness*
- *patient centredness*
- *timeliness*
- *efficiency*
- *equity'*

'For your project can you think of something that you feel could be done better? For example the open access arrangements and plans for patients with chronic medical conditions? The effectiveness and timeliness of handover ... etc.'

Draw a diagram of the PDSA cycle (Plan-Do-Study-Act).

Closing remarks

'Can I summarise what we've talked about. Firstly, tell me what you understand makes up a quality improvement project?'

'Remember that your ultimate aim is to improve the quality of patient care. There is a department that can help you with your project within this trust. If you are struggling to come up with an idea we could discuss this again in a few days. You should also talk to your educational supervisor about how you are planning on carrying out this project and your time frames. Do you have any other questions? Do you feel clearer about how you would go about starting your project?'

Background notes

Quality Improvement and Patient Safety (QIPS)

The aim is to improve the service we provide by making it more safe, efficient, effective or in some other way improving the patient's experience:

1. Identify a project:
   - ○ This is often the main stumbling block, particularly for a very junior doctor who may not necessarily recognise when things aren't working as well as they could, having nothing to compare to:
     - Get ideas from more senior members of the team if necessary
2. Decide what change you want to make:
   - ○ Start small, you can carry out many cycles of change
3. Engage stakeholders:
   - ○ ie talk to everyone who is involved, explaining the change you are proposing; there is no way the change will be made and will be sustained without this!

4. Make the change
5. Study the effects of the change:
   - eg audit length of handover/survey junior doctors' opinions
6. Make further changes that may have arisen to continue improvement.

There is a lot of helpful information about quality improvement on the RCPCPH website in the 'Quality Improvement and Clinical Audit' section.

5.4 Case four: obtaining consent for a blood transfusion

Candidate information

Background

You are a neonatal Specialist Trainee. You are looking after a preterm infant called Toby Gibson. Toby was born at 26 weeks and is now 4 days old. The baby had initial problems with respiratory distress syndrome and hypotension requiring inotropes. You note that he is becoming anaemic and suspect this is because of frequent sampling from his umbilical arterial line for gases and bloods. You feel he would benefit from a transfusion. You have arranged to talk to his mother about this.

Task

Explain the reasons for the blood transfusion and obtain consent.

Role player information

○ This is your first child.
○ You have been very worried about Toby for the last few days.
○ You are anxious about the risks of blood transfusions.

Questions you should ask

○ Does he really need to have a transfusion; what would happen if he didn't?
○ Can you be sure that he won't get any infections from the blood?

Questions you could ask

○ Can I give him my blood for his transfusion?
○ Will the transfusion make him better?

Examiner information

Guide notes towards Expected Standard

○ Acknowledges parental concerns
○ Explains indication for transfusion
○ Explains expected benefit
○ Explains risks of transfusion (wrong blood/wrong patient/transmission of bacterial or viral infection).

Suggested plan of attack

Preparation

Make short notes about which points you need to cover. These could look like this:

1. Reason for anaemia (frequent sampling, immature bone marrow, intraventricular haemorrhage).
2. Reason for transfusion (oxygen requirement, hypotension, Hb unlikely to recover spontaneously in next few weeks).
3. Risks of transfusion and safety checks (mislabelling/wrong blood/infection checks on donated blood).

Introduction

Introduce yourself and explain where you fit into the team looking after Toby. Check parents' understanding of Toby's current situation.

> *'Hello Mrs Gibson, my name is Dr Goodwill. I am the specialist trainee looking after Toby. I need to talk to you about some of the plans we have for Toby's treatment.'*
>
> *'Before we start, could I just check what you understand about Toby's current situation?'*

Talk about anaemia and the negative impact on Toby's ventilation and blood pressure management.

Explanation

- ○ How transfusion will help
- ○ Need for consent
- ○ Opportunity for questions
- ○ Talk through risks and explain what is done to minimise these.

Closing remarks

Need to summarise what you've said and check that the parent understands.

> *'May I summarise what we've talked through? I've told you that Toby is stable but he is anaemic. This is a very common problem we see in babies born prematurely. Unless he has a transfusion we expect this will affect his breathing and his blood pressure. We have talked about the risks of transfusion and how these are small and every effort is made to minimise them with procedures for checking donors and double checks to make sure the right blood is given to the right baby. Have you got any further questions about anything I've said?'*
>
> *'Do we have your permission to give Toby a transfusion?'*

Notes on consent

- Consent can be obtained from someone with parental responsibility
- Only one person with parental responsibility is needed, although ideally all parties with parental responsibility should consent
- The child's birth mother always has parental responsibility unless the child has been adopted
- The child's natural father has parental responsibility if they were married at the time of the child's birth, or if his name is on the birth certificate, or if he has acquired parental responsibility through the courts (residence order)
- A step-parent can consent if they have undergone a court process to assume parental responsibility
- If a child is the subject of a Care Order then social services can consent on behalf of the local authority
- An Emergency Protection Order gives the local authority the ability to exercise parental responsibility if it is reasonably required to safeguard or promote the welfare of the child
- A foster parent does not have parental responsibility
- A child can consent for themselves if they are aged 16–18. This autonomy can be suspended by the courts in those under 18 years old where required
- At any age a child can consent to treatment if they understand the issues involved; this is called 'capacity' (also 'Gillick competence'). The lower limit of Gillick competence age is related to the child's experience and the gravity of the treatment for which they are consenting. They can agree to treatment but cannot refuse treatment if they are under 16.

What to do if parents disagree with you

This depends on degree of urgency. You can act to give treatment without parental consent if an immediate medical procedure is required to save life or prevent irreversible harm. In less urgent cases where there is a difference of opinion between medical staff and parents about what is in the child's best interests, the following is a potential stepwise approach:

1. Consult local colleagues (eg senior colleagues/other consultants).
2. Consult more widely within the hospital (involve hospital ethics committee).
3. Consult another unit.
4. Involve Royal College ethics committee.
5. Involve the courts: a judge can issue a Specific Treatment Order. This gives permission to act without the parent's consent in one particular area but does not take away any other aspects of parental responsibility.

In cases where parents are insisting on treatment that medical staff do not think is in the child's best interests, a similar process can be followed. As a doctor you do not have to do anything that you think is wrong, even if the parents are insisting (it is the child who is your patient). You are required to supply a second opinion when requested to do so.

Risks of transfusion

Highest risks occur when blood is being given out of hours. Reactions are most likely to occur within the first 15 min of transfusion; therefore start slowly for 15 min. Because there is a risk associated with transfusion, it is very important that the indications for transfusion are clearly documented, and that the discussion asking for consent is clearly recorded also. Most hospitals have a policy of giving written information about transfusions to patients or their parents.

The main risks are as follows:

1. Wrong blood/wrong patient
2. Haemolysis
3. Bacterial contamination
4. Anaphylaxis/allergy
5. Transfusion-associated acute lung injury
6. Risk of infection:
   - Hepatitis B virus (HBV) – 1:900 000
   - Hepatitis C virus (HCV) – 1:30 million
   - HIV – < 1:several million
   - Variant Creutzfeldt–Jakob's disease (vCJD) – one possible case of transmission so far; potential for existence of transfusion-acquired vCJD still under debate. vCJD remains a rare condition in the UK (145 cases to date) but future prevalence is unknown, so several precautions are in place (particularly for plasma-related products).

Jehovah's witnesses

- Jehovah's witnesses do not accept transfusion of whole blood, red cells, platelets, white cells or plasma; other 'fractions' of blood (for example clotting factors) are left to the believer's own conscience.
- This is based primarily on the interpretation of Bible passages, backed up by knowledge of the risks and alternatives to transfusion.
- If an adult accepts a transfusion they risk excommunication. If their child is forced to have a transfusion by the courts this does not apply, yet the perceived spiritual consequences for their child will be distressing for their family irrespective of a court order.

The following website and paper may be helpful when addressing the issue of blood transfusions in Jehovah's witnesses:

- www.transfusionguidelines.org.uk
- http://adc.bmj.com/content/90/7/715

Some useful websites:

- www.blood.co.uk (National Blood Service website)
- www.shotuk.org (Serious Hazards of Transfusion website)

5.5 Case five: Duty of Candour – wrong antibiotic prescribed

Candidate information

Background

You are a Specialist Trainee in paediatrics. The ward sister has just told you that a patient who is allergic to penicillin has been given two doses of iv penicillin in the last 6 hours. One of the student nurses noticed that on the front of the drug chart it was written that he was allergic to penicillin – this was also documented in the admission clerking. The night registrar had prescribed the iv penicillin to treat cellulitis. The boy had no apparent reaction to either of the doses.

Task

Explain to the mother what has happened, and why you are telling her this information.

Role player information

You are the mother of Sebastian. You have three other young children at home and didn't get much sleep last night while staying on the ward with him. You are feeling tired and worried, and just want to get home, but your priority is what's best for your son.

Questions you should ask

○ Does this mean Sebastian is not allergic to penicillin?
○ What could have happened to him if he had been allergic and had been given the penicillin?
○ What's the point in going through the story so many times to so many people when no one listens to a word you're saying?
○ Why are you even bothering me with this information?

Examiner information

Guide notes towards Expected Standard

○ Clearly and confidently explains the situation
○ Apologises for the mistake
○ Discusses how this error occurred and that this will be investigated further
○ Explains what could have happened if asked (ie anaphylaxis)
○ Explains why this information is being shared
○ End with reassurance and a further apology.

Suggested plan of attack

Introduction

Introduce yourself and explain your role in Sebastian's care. Explain why you have come to talk to her.

> *'Good afternoon Mrs Jones. My name is Dr McGlone, I'm one of the paediatric registrars who is looking after Sebastian. I have come to talk to you about the antibiotics Sebastian has been having for his skin infection. Sebastian has had two doses of intravenous penicillin. The reason why I have come to talk to you about this is that this prescription was written in error as you had informed us that Sebastian was allergic to penicillin ...'*

Explanation

After quickly getting to the point, you can explain that we as medical health professionals do not keep anything from patients, and that we have a duty and a responsibility to be open and honest. It is very fortunate that in this situation no harm has come to Sebastian – however it could have, and because of this, further action will be taken to try and reduce the chance of this ever happening again. It is also important that Sebastian and parents know that he is not allergic to penicillin. You can end with:

> *'All the staff on the ward are extremely sorry that this has happened. I would like to reassure you that no harm has come to Sebastian; we will continue to give him penicillin as it is the most effective antibiotic in this condition. Do you have any questions that I could answer for you?'*

Key Documents

- ○ *Medico-legal Guide to Statutory Duty of Candour* (www.themdu.com)
- ○ *Introducing the Statutory Duty of Candour* (www.gov.uk)

5.6 Case six: talking about a chronic condition – poorly controlled diabetes

Candidate information

Background

You are a Specialist Trainee working in a paediatric diabetes clinic. Kieran is a 15-year-old boy with diabetes. He initially had good control with HbA1cs between 53 and 63 mmol/mol (7.2%–7.9%) until the last 18 months. His most recent HbA1c was 97 mmol/mol (11%). He was recently admitted after being found unconscious after a big night out when he admits he was drinking alcohol. It was felt he had had a hypoglycaemic seizure. He has not brought his blood glucose meter with him, but says he hasn't checked any blood sugars since his last appointment. You are seeing him on his own – without his parents.

You do not need to take any more medical history.

Task

Talk to Kieran about improving his glycaemic control, and the effect alcohol will have on his blood glucose levels.

Role player information

You have been diabetic since you were 6 years old. You are having constant arguments with your parents because they say you are not taking enough care of your own diabetes. You don't tend to check your blood sugars unless you are feeling unwell.

Your general feelings

○ People who don't have diabetes don't understand what it is like:
  - You hate having diabetes and just want to be a normal teenager and do all the things your friends do

○ Being admitted to hospital was frightening

○ You do not want to check your glucose levels at school because you are worried about what your friends will say.

Examiner information

Guide notes towards Expected Standard

○ Appropriate conduct of interview

○ Invites Kieran to share his own concerns

○ Explains need for more frequent blood glucose testing:
  - Doesn't lecture about alcohol but gives appropriate advice regarding drinking safely with diabetes

○ Agrees a realistic plan of action

Suggested plan of attack

Preparation

This is a non-compliant teenager and an example of a scenario where you need to explore his attitude towards his condition. Similar issues arise in many adolescent patients with chronic conditions. There are a variety of different models or styles of consultation. In the poor compliance scenario, motivational interviewing is often used (www.motivationalinterviewing.org). Points to consider when you are faced with a non-compliant teenager are listed in the Box.

With these points in mind, during your preparation you could list some key points to discuss. These could include:

1. Establish agreed agenda for consultation: invite him to express his concerns.
2. Main difficulties for Kieran.
3. Outline problems (high HbA1c, need for more monitoring, hypoglycaemic seizure – the implications re: driving and general safety).
4. Outline health concerns if no change made (ketoacidosis/hypoglycaemia/long-term problems).
5. Action plan before next clinic.
6. Offer extra support/bring forward next clinic appointment/specialist nurse appointment/home visit.

Points to think about when you are faced with a non-compliant teenager

1. Never assume they know everything they need to about their condition. Often the reasons behind the treatment are explained to the parents at diagnosis and then not revisited. The patient may have been too young to understand or may not remember being told some essential information. Make sure they are clear about the facts.
2. Find something positive to say.
3. Show respect and empathy – it's difficult for them to have this condition.
4. Be honest about what you need from the consultation.
5. Establish roles in the management of the condition. Define explicitly who has responsibility for which aspect of management.
6. Ask the patient what their main concerns are and whether they can see any solutions for those. Ask them how they would like help.
7. Try to agree on a way forward. Start from where they are at. Short-term goals are best. Aim for one small step at a time. Aim for goals that are achievable.
8. Explore support network. Think of ways they could include their peers in their support network. Remember that specialised support groups exist and are sometimes extremely helpful.
9. Think about how your multidisciplinary team could help.
10. Consider psychological or psychiatric co-morbidities: suicidal ideation, risk-taking behaviour, bullying, family stressors, self-harm and depression.

Introduction

Need to establish common ground and a common agenda.

> *'Good morning Kieran, my name is Dr Goodwill. I am the specialist trainee at the diabetes clinic. It's great that you've come to clinic today. How are you? … How are things going with your diabetes?' Allow time for him to answer, we can feel uncomfortable with silence but this gives the patient some space to think about what they want to talk about.'*
>
> *'Was there anything in particular about your diabetes that you wanted to talk about?'*
>
> *'There are two things I think we need to discuss. The first is about your recent hospital admission and the effect alcohol has on your blood sugars, and the second is about checking your blood sugars more regularly.'*

> *'Do you understand what happened around the time you were admitted to hospital, and what led up to that? You said at the time you'd had quite a bit to drink – maybe now would be a good time to talk about alcohol and how it affects your blood glucose levels. Do you know what effect it has?'*
>
> *'Secondly I'm concerned that you haven't brought your meter with you today. Have there been any problems with checking your sugars recently?'*
>
> *'Do you think you should check your sugars more often; do you think this would help with control of your diabetes?'*

Explanation

> *'Your HbA1c has been high for a while now which means your blood glucose levels will be high most of the time; this could mean that the insulin you are on is not right for you. It's difficult for us to know if we need to change things if we can't see what is happening to your sugars.'*
>
> *'Any tests are better than no information. Let's set two goals for you to achieve between now and when we next meet. What would you like your goals to be ?'*

Closing remarks

Review what you've spoken about. Reiterate your agreed action plan and check that he has understood. Make space for any extra questions. Offer extra support; this could be a support group for children with chronic conditions, adolescent expert patients programme, or further support available through the diabetes multidisciplinary team (specialist nurse follow-up phone call, appointment with team psychologist).

> *'Kieran, thanks for being so honest with me about how diabetes is affecting things for you. We have talked about your diabetes, the effect of alcohol and how blood testing helps monitor how well your diabetes is controlled.'*
>
> *'The goals you have set yourself for your next appointment are: ... Have you got any questions about anything we've talked about today?'*

5.7 Case seven: giving information – telephone conversation about hydronephrosis

Candidate information

Background

You are a Specialist Trainee in paediatrics, working in a district general hospital. You will be talking to Mrs Matthews, mother of Charlie, who is a 3-month-old baby. Charlie was admitted 2 weeks ago with a urinary tract infection. He was discharged after 2 days and an outpatient renal ultrasound was arranged. The report shows significant right-sided hydronephrosis suspicious of reflux.

On the ward Charlie was given intravenous antibiotics and responded quickly. He was seen at the end of his course of antibiotics and had made a full recovery. Mrs Matthews is expecting to be contacted with the results of the ultrasound. She was told that, if it was normal, no further follow-up would be arranged.

Task

Talk to Mrs Matthews about the ultrasound result and what your further management will be.

Role player information

You are Mrs Matthews, mother of Charlie, who is a 3-month-old baby. He is your first child. Two weeks ago you took Charlie to see your GP because he had not been feeding well for 2 days and had developed a high fever. He was referred to hospital and immediately put on intravenous antibiotics for what you later found out was a urinary tract infection. You were relieved that he responded quickly to the antibiotics, and after 2 days he was switched to oral antibiotics and sent home. At the end of the course of oral antibiotics he was seen again at the hospital and you were told that he had made a full recovery, but that he would need an ultrasound scan of his kidneys because he was very young to have had a urinary tract infection, and sometimes this can mean that there is something wrong with the kidneys. You took him for his scan earlier in the day and you are expecting the hospital team to ring you with the results.

Your general feelings

○ You feel guilty that you did not take Charlie to hospital sooner. He had been unwell for 2 days before you took him to the GP, before his being diagnosed with a urinary tract infection.

- ❍ You are very anxious that the ultrasound scan will show something very serious.
- ❍ You are worried that you will not notice if Charlie becomes unwell again, and this may mean that his kidneys become damaged.

Questions you should ask

- ❍ Is it something serious?
- ❍ Is it my fault because I didn't bring him to hospital soon enough when he had a urinary tract infection?
- ❍ What are the symptoms of urinary tract infections in babies? How will I be able to tell if Charlie is becoming unwell again?

Examiner information

Guide notes towards Expected Standard

- ❍ Provides an appropriate, clear greeting and clarifies the purpose of the call
- ❍ Explains clearly the abnormality on the ultrasound scan
- ❍ Appropriately addresses parental concerns
- ❍ Provides an explanation of the symptoms of urinary tract infections and when it is necessary to seek help
- ❍ Checks Mrs Matthews' understanding of what was discussed

Suggested plan of attack

Preparation

You are phoning with an abnormal result. Although it is less serious than some potential diagnoses, you need to be prepared for this to be a 'breaking bad news' situation because you cannot be sure how the parents will react. You should follow a similar pattern to that for any scenario which involves breaking bad news.

Make short notes about the key points you need to discuss. These could include the following:

- ❍ Establish how much the mother already knows
- ❍ Introduce new information – scan result, probable diagnosis of vesico-ureteric reflux (VUR)
- ❍ Provide an explanation of VUR
- ❍ Give the mother an opportunity to ask questions
- ❍ Explain what happens next – organise micturating cystourethrogram (MCUG), start prophylactic antibiotics, arrange clinic follow-up
- ❍ Describe how to watch for signs of a UTI.

Introduction

How you start the conversation will be critical to how the examiner interprets your 'conduct of interview'. Because your body language and ability to pick up non-verbal cues are not being assessed, the clarity of your initial greeting will carry much more weight.

> *'Hello, my name is Dr Smith. I'm a paediatric registrar from City Hospital. I would like to talk to Charlie Matthews' mother.'*
>
> *'Is now a good time to talk about Charlie's ultrasound scan result?'*
>
> *'What do you understand about why the scan was done?'*

Explanation

Present the new information.

> *'The ultrasound scan showed that one of Charlie's kidneys has an abnormality. There was a fluid-filled enlargement inside the kidney which is called hydronephrosis.'*

Explain what it means.

> *'Normally this is caused when a valve in the bladder is immature and allows urine to flow back up towards the kidneys when the bladder empties. This is called reflux, and it causes the sort of abnormality that we can see on Charlie's kidney scan.'*
>
> *'In most cases the abnormality goes away as the child gets older, and does not cause lasting damage. However, as long as it is there Charlie will be at increased risk of getting urinary infections, which is why it is important that we know about it and why we arranged for him to have the scan.'*

Provide an opportunity for questions.

> *'Have you any questions about anything I have said so far?'*

Explain what happens next.

> *'What we need to do next is to arrange another scan called an MCUG. This involves putting a small tube called a catheter into Charlie's bladder, putting some fluid into it which will show up on X-ray, and looking to see if the urine is going back up towards the kidneys. I will also arrange for Charlie to come back to clinic to see a paediatrician a few weeks after he has had the scan, so that we can see how he is and talk about the results. In the meantime, Charlie will need to be given some antibiotics to prevent him getting more urinary tract infections. I will ask your GP to prescribe these.'*

Provide a further opportunity for questions.

> *'Have you any questions about the scan?'*

Explain the symptoms of a UTI and when it is necessary to seek help.

> *'Even with preventative antibiotic, breakthrough infections can still occur. It is very important that you see a doctor quickly if you are concerned that Charlie is unwell and there isn't another obvious cause. This is particularly important for Charlie, because if he were to have an infection in his bladder, this infected urine could go towards his kidneys and cause a more serious kidney infection. The sort of symptoms that babies have when they have a urinary infection are fever without an obvious cause, strong-smelling or offensive-smelling urine, not feeding very well, being irritable or vomiting a lot.'*

Closing remarks

Summarise what you have said and check the mother's understanding.

> *'Can I summarise what we have talked through? The scan shows that Charlie may have reflux from his bladder to his kidneys. I will arrange for another scan to see if this is the case, and talk to your GP about getting Charlie some antibiotics to be given between now and when he next comes to clinic. In the meantime, you should take him to the doctor if he has any symptoms that could be a urinary tract infection. Have you any questions about anything I have said?'*

Explain the arrangements for documentation and follow-up.

> *'I will send you a letter summarising what we have talked about, along with some written information about urinary tract infections in babies and the MCUG scan. I will also send a copy to your GP. In addition, you will receive an appointment for the MCUG scan and another one for Charlie's clinic follow-up. Please contact us if you have any questions in the meantime.'*

Background notes

Urinary tract infections

Guidelines on the investigation and treatment of urinary tract infections in children were published by NICE in 2007 (www.nice.org.uk/cg54). The guidelines specify which investigations are recommended for urinary tract infection, depending on the age and presentation of the child.

Vesico-ureteric reflux (VUR)

- This is the retrograde flow of urine from the bladder to the kidneys

- It can be primary (in an otherwise normal renal tract) or secondary (eg in a neurogenic bladder or with posterior urethral valves)
- It is the most common abnormality in children who have UTIs
- It is classified according to five grades of severity, ranging from grade 1 (mild) to grade 5 (severe, with damage to the renal parenchyma)
- Around 80% of cases of mild to moderate VUR will completely resolve by the age of 5 years. However, severe reflux is unlikely to resolve spontaneously
- There is a risk of renal scarring if infected urine refluxes from the bladder to the kidneys, causing pyelonephritis. There is also a risk of reflux nephropathy
- Around 20% of cases with scarring on DMSA develop hypertension later in life (increased renin levels because of damaged renal tissue)
- Treatment involves prompt recognition and treatment of UTIs (some children require prophylactic antibiotics), surveillance of renal growth and function, monitoring of blood pressure if complications are identified and treatment of associated conditions that can predispose to UTIs (eg dysfunctional voiding, constipation)
- Surgical correction is rarely necessary, but is indicated for severe reflux that is unlikely to resolve, worsening renal function, frequent breakthrough pyelonephritis or persistent reflux in older children.

6

FURTHER SCENARIOS

There now follow suggested scenarios to help direct your revision. These are all based on topics that have come up in the exam before. They are intended to be used as a starting point for you to practise adapting your structure and technique for this section of the exam to a range of different topics. Some background information is provided although this is not extensive and as with the scenarios previously, gives guidance rather than precise answers.

Practise in pairs with a facilitator where possible. Actually role play the scenario rather than saying 'I would say this'.

Don't panic if you look at one of these scenarios and think you don't know anything about the topic. The communication stations are definitely about conveying concrete information as well as how you go about doing this. In reality we can't all know everything about everything. In the exam take a deep breath, keep calm and think about your approach. You can say – 'I'm afraid I don't have that much experience with this particular condition, but I can certainly find out for you – let's talk through your questions/concerns, then I can get some specific information for you for when we meet again tomorrow'... or next week ... or whenever you feel is appropriate.

1. **A child has a congenital myopathy and is ventilator dependent on the neonatal unit. Her parents wish for a withdrawal of intensive care. Talk to a medical student about the ethics of withdrawing intensive care in this case**
   - *Making Decisions to Limit Treatment in Life-limiting and Life-threatening Conditions in Children: A Framework for Practice* – this document is accessible from the RCPCH website (see below) and lists three situations when limiting treatment can be considered as it may not be in the child's best interest to continue:
     - **When life is limited in quantity** (ie treatment is not likely to prolong life significantly)

  - **When life is limited in quality** (where treatment may prolong life significantly but will not alleviate the burdens associated with the illness or treatment)
  - **Informed competent refusal of treatment** (only applies to older children who repeatedly and competently consent to withdrawal of treatment)
- This will be a team decision involving the multidisciplinary team involved and family members
- Mention palliative and end-of-life care. Suggest they go away and spend some time looking at guidance available:
  - *Making Decisions to Limit Treatment in Life-limiting and Life-threatening Conditions in Children: A Framework for Practice* (adc.bmj.com)
  - Palliative care on neonatal units (www.togetherforshortlives.org.uk)

2. **Talk to a medical student about the ethics of offering neonatal intensive care to a child born at 23 weeks**
   - Make sure you are aware of the up-to-date survival data and long-term morbidity of infants born at this gestation (www.epicure.ac.uk)
   - Autonomy – consider the parents' right to decide, giving the child a chance of an autonomous life
   - Beneficence/non-maleficience – think about the impact on the family of the ups and downs of neonatal intensive care, and of looking after a potentially disabled child; for some people this would be a significant 'harm' and for others it would be a 'good'
   - Justice – NICU is expensive, beds in tertiary units may be scarce
   - Remember to seek the medical student's own personal views and experiences and discuss these too
   - In general, a senior neonatologist will make this decision, taking the parents' view into account

3. **'Colleague in difficulty' scenario**
   - Talk to a Specialist Trainee who has been refusing to help clerk-in general paediatric patients at busy times
   - Talk to a Specialist Trainee who is refusing to work the shift he has been allocated on New Year's Eve
   - Talk to an F2 who has not completed any discharge summaries for several months. Other juniors are complaining that he does not pull his weight:
     - These are common communication station scenarios and you would approach them in a similar way
     - Mention that there have been concerns regarding ... be specific

6

FURTHER SCENARIOS

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Don't panic if you look at one of these scenarios and think you don't know anything about the topic. The communication stations are definitely about conveying concrete information as well as how you go about doing this. In reality we can't all know everything about everything. In the exam take a deep breath, keep calm and think about your approach. You can say – 'I'm afraid I don't have that much experience with this particular condition, but I can certainly find out for you – let's talk through your questions/concerns, then I can get some specific information for you for when we meet again tomorrow'... or next week ... or whenever you feel is appropriate.

1. **A child has a congenital myopathy and is ventilator dependent on the neonatal unit. Her parents wish for a withdrawal of intensive care. Talk to a medical student about the ethics of withdrawing intensive care in this case**
   - *Making Decisions to Limit Treatment in Life-limiting and Life-threatening Conditions in Children: A Framework for Practice* – this document is accessible from the RCPCH website (see below) and lists three situations when limiting treatment can be considered as it may not be in the child's best interest to continue:
     - **When life is limited in quantity** (ie treatment is not likely to prolong life significantly)

  - **When life is limited in quality** (where treatment may prolong life significantly but will not alleviate the burdens associated with the illness or treatment)
  - **Informed competent refusal of treatment** (only applies to older children who repeatedly and competently consent to withdrawal of treatment)
- ○ This will be a team decision involving the multidisciplinary team involved and family members
- ○ Mention palliative and end-of-life care. Suggest they go away and spend some time looking at guidance available:
  - *Making Decisions to Limit Treatment in Life-limiting and Life-threatening Conditions in Children: A Framework for Practice* (adc.bmj.com)
  - Palliative care on neonatal units (www.togetherforshortlives.org.uk)

2. **Talk to a medical student about the ethics of offering neonatal intensive care to a child born at 23 weeks**
   - ○ Make sure you are aware of the up-to-date survival data and long-term morbidity of infants born at this gestation (www.epicure.ac.uk)
   - ○ Autonomy – consider the parents' right to decide, giving the child a chance of an autonomous life
   - ○ Beneficence/non-maleficience – think about the impact on the family of the ups and downs of neonatal intensive care, and of looking after a potentially disabled child; for some people this would be a significant 'harm' and for others it would be a 'good'
   - ○ Justice – NICU is expensive, beds in tertiary units may be scarce
   - ○ Remember to seek the medical student's own personal views and experiences and discuss these too
   - ○ In general, a senior neonatologist will make this decision, taking the parents' view into account

3. 'Colleague in difficulty' scenario
   - ○ Talk to a Specialist Trainee who has been refusing to help clerk-in general paediatric patients at busy times
   - ○ Talk to a Specialist Trainee who is refusing to work the shift he has been allocated on New Year's Eve
   - ○ Talk to an F2 who has not completed any discharge summaries for several months. Other juniors are complaining that he does not pull his weight:
     - These are common communication station scenarios and you would approach them in a similar way
     - Mention that there have been concerns regarding ... be specific

- 'How are you – how are you finding this job?' (it may be a personal problem that they need to be supported with, or a specific anxiety about/within the job)
- Try to understand why they are doing this and come up with a solution
- Be clear about expectations
- 'Do you think that what I am saying is fair?'
- Make a plan as to how you are going to move forward
- Arrange to meet a week or two later to see how they are getting on
- Know how and who to escalate to if required.

4. **Talk to a medical student about why we use some medications that are unlicensed for children**
   - ○ Licensing refers to a drug being given marketing approval to be advertised for a specific clinical situation
   - ○ Pharmaceutical companies perform Phase 1, 2 and 3 trials before a licensing application:
     - Phase 1 trial = a few selected patients or healthy volunteers (looks at safety, pharmacokinetics, etc)
     - Phase 2 trial = a few more patients (looks at best dose and whether the medication works)
     - Phase 3 trial = comparison with best existing medication (randomised controlled trial with larger numbers)
     - Phase 4 trial = after a drug has been made widely available (post-licensing)
   - ○ It is often felt to be unethical to perform randomised controlled trials on children, especially if a medicine clearly works in adults
   - ○ Newer medications take a very long time to become widely used in paediatrics because of this (and therefore have more years of 'adult' experience prior to their being used)
   - ○ There are some excellent sources of information about which unlicensed medications are acceptable and how these are generally used in paediatrics (*Medicines for Children* available on the Medicines and Healthcare products Regulatory Agency website at www.mhra.gov.uk, and the *BNF for Children*, available online at http://bnfc.org)
   - ○ New European Union legislation was introduced in January 2007 requiring pharmaceutical companies to make a plan for investigating whether their medicines can become available for children when they register these medicines for adults. This is called a Paediatric Investigation Plan and is monitored by the European Medicines Agency. The Medicines for Children Research Network (MCRN) is a good source of further information (www.mcrn.org.uk)

5. **Obtain consent for insertion of an umbilical arterial catheter (UAC)**
   - ○ Indications/Benefits – blood pressure monitoring, blood sampling
   - ○ Explain what the procedure entails
   - ○ Risks – failure to get the UAC in, blood loss, accidental displacement, line infection, necrotising enterocolitis, arterial thrombosis (particularly renal, distal ischaemia, eg in limbs)
   - ○ Complications avoided by checking for correct placement, close monitoring for complications and removing the UAC as soon as it is no longer needed, or if any suggestion of any complication.

6. **Talk to a mother who is refusing to allow an F2 to insert a cannula into a well 2-year-old child with a petechial rash**
   - ○ Explain why the procedure is being done, and what the likely benefit is (able to take blood for diagnosis, able to give antibiotics and fluids if required should child deteriorate suddenly)
   - ○ Explain the risks of the procedure (failed attempts, pain and distress to child) and what will be done to minimise these (topical analgesia, distraction, experienced medical and nursing staff)
   - ○ Explore alternative options available
   - ○ Explore reasons why mother is resistant

7. **Tell the parents of a 3-year-old that their child was accidentally given a double dose of gentamicin**
   - ○ Apologise for the mistake
   - ○ Main risks: renal toxicity (often reversible), ototoxicity (tinnitus, ataxia), which can be permanent
   - ○ What to do for the patient – check gentamicin levels, renal function, hearing if levels are high, monitor urine output and blood pressure
   - ○ What you are going to do to avoid another mistake – incident form, staff education, discussion at governance meeting.

8. **Talk to the mother of a premature baby on the neonatal unit who has had a skin swab come back positive for MRSA. He has previously had negative swabs**
   - ○ Apologise for the situation (this is not accepting blame as in this case the MRSA may not have been transferred by NHS workers – up to 3% of the population are colonised with MRSA)
   - ○ Establish mother's current understanding of what MRSA is
   - ○ Inform her that it lives on the skin without causing symptoms, many people are 'carriers' without knowing it
   - ○ Explain that if it causes an infection this can be more difficult to treat
   - ○ Explain the baby will need to be isolated and depending on the unit's guidelines, will almost certainly have topical treatment administered
   - ○ The baby will have repeat swabs after treatment to ascertain whether treatment has been successful

9. **Discuss the diagnosis of diabetes with the mother of a 5-year-old admitted with weight loss, polyuria, polydipsia and glycosuria**
   - ○ 'How much do you know about diabetes? Do you know anyone who has diabetes?'
   - ○ Stress that nothing could have been done to cause or prevent this from happening
   - ○ Lifelong condition, not curable but is treatable
   - ○ Need for insulin injections, glucose monitoring
   - ○ Referral to diabetes team for more verbal and written information.

10. **Talk to the parents of a child who was unexpectedly found to have ambiguous genitalia**
    - ○ Congratulations first on their new baby
    - ○ Talk about the unusual appearance of the external genitalia; 'Need to obtain more information before assigning a gender'
    - ○ Do not register the birth yet
    - ○ Referral to specialist multidisciplinary team (endocrinologist, urologist, geneticist, psychologist)
    - ○ Need for tests (karyotype, pelvic ultrasound scan, pituitary hormones, 17-hydroxyprogesterone)
    - ○ Stay in hospital: monitor blood glucose/blood pressure/urea and electrolytes until congenital adrenal hyperplasia is excluded.

11. **Discuss with the mother of a 25-week baby who has periventricular cysts on routine ultrasound scan at 20 days of life. The baby had not previously had IVH found on scans**
    - ○ Explain why preterm babies can develop these cysts – fluctuations in blood flow to areas of the brain – associated with changes in blood pressure/sepsis
    - ○ Long-term implications with development – be guarded, it is likely it will be affected but impossible to predict to what extent
    - ○ Baby will be followed up through infancy and into childhood to detect and manage any developmental difficulties.

12. **Talk about starting inhaled steroids with the mother of a 5-year-old who has had recurrent wheeze and who has had four courses of oral steroids in the last 12 months; she is concerned about effects on growth**
    - ○ Establish common ground: what does she know about inhaled steroids, what are her main concerns?
    - ○ Explain that poorly controlled asthma has significant effects on growth
    - ○ Talk about bioavailability/drug delivery and how these lessen systemic effects
    - ○ Reassure and emphasise the benefits of improved control
    - ○ Make a plan for reassessing the need for steroids depending on response

13. **Talk to a 16-year-old girl with diabetes who is anxious about being transferred to adult services**
    - ○ Explore specific causes for concern
    - ○ Discuss transition clinics – similarities/differences
    - ○ Explain where they can go for further support (expert patients programme/Diabetes UK (www.diabetes.org.uk)/ adult diabetes specialist nurse)
    - ○ Reassure and emphasise positive aspects.

14. **Talk to a 14-year-old girl with cystic fibrosis about the need for (her first) admission to hospital for iv antibiotics**
    - ○ Explain the need for treatment in hospital – frequent iv antibiotics/ intensive physiotherapy input/aim to increase lung function
    - ○ Explain what will be involved – staying in a cubicle/insertion of a long line
    - ○ Explore anxieties/main perceived difficulties
    - ○ Emphasise benefits.

15. **The GP trainee on your team finds a heart murmur in a baby on the postnatal wards. Talk to the parents**
    - ○ Establish common ground – what do they know about heart murmurs?
    - ○ Explain what a murmur is and potential causes for this (innocent/from closing duct/hole or narrowing in heart or blood vessels coming from heart)
    - ○ Explain what will happen next – four-limb blood pressure and saturations ECG/echo +/– cardiology referral.

16. **Talk about contraceptive choices with a sexually active 15-year-old girl who has epilepsy**
    - ○ Check that no abusive situation is occurring, check she knows the law relating to consent
    - ○ Check that she knows about safe sex and the risks and consequences of sexually transmitted diseases
    - ○ Explain that anti-epileptics can interfere with the combined oral contraceptive pill, therefore she will need a higher than normal dose
    - ○ Explain that the progesterone-only pill doesn't work in these circumstances
    - ○ Explain that depot injections don't last as long (needed every 10 weeks)
    - ○ Explain that a higher dose is needed for emergency contraception
    - ○ Explain where to go for further help (family planning clinic/general practitioner/specialist nurse/practice nurse/websites)
    - ○ Explain that some anti-epileptic drugs can cause harm to an unborn baby, as can uncontrolled epilepsy in pregnancy

- ○ Explain that she will need to take higher than usual doses of folate when planning to become pregnant.

17. **Explain the inheritance of Prader–Willi syndrome to a medical student**
    - ○ Check prior knowledge – of imprinting
    - ○ Describe Prader–Willi in terms of having 'no active paternal Prader–Willi gene'
    - ○ Could be a new mutation in the child affecting the chromosome 15 that was inherited from the father
    - ○ Could also occur from uniparental disomy (25% of cases) – the child receives both chromosomes 15 from the mother (and so lacks a paternal copy of the gene)
    - ○ Use diagrams.

18. **Talk to the parents of a child with Down syndrome about the likelihood of future children having the condition**
    - ○ Check prior knowledge about chromosome inheritance in general and trisomy 21 in particular
    - ○ Best discussed using diagrams
    - ○ Down's occurs because of meiotic nondisjunction in 94% of babies
    - ○ For most families where one child has had confirmed trisomy 21 there is a 1 in 100 chance of having a baby with another chromosomal abnormality
    - ○ If the mother is over 35 years old, the risk of recurrence is double the baseline risk for age (increases with increasing age)
    - ○ Translocation (onto another chromosome, 15, 22 or 21) occurs in 5% of babies – in a quarter of these one parent carries a balanced translocation
    - ○ The risk of recurrence is 10%–15% if the mother has a translocation and 2.5% if the father has a translocation (or 100% if the translocation is 21:21 – rare)
    - ○ Mosaicism occurs in 1% of babies with Down's – trisomy arises post-fertilisation during mitosis.

19. **This 15-year-old boy has recently been diagnosed with epilepsy. Talk about how this will affect his lifestyle**
    - ○ Advise him to tell anyone with whom he spends a fair amount of time what may happen and what to do if it does
    - ○ Advise him to think about what would happen if he had a seizure in different situations
    - ○ Tell him that showers are safer than baths, and to use a sign rather than locking the bathroom door
    - ○ Tell him to take commonsense precautions for sport activities
    - ○ Advise him that this will affect driving if it is not controlled by then (stress the need for compliance with treatment) and that he can only

apply for a license if he is seizure free for 12 months (or night-time only seizures for 3 years)
- ○ Advise him that anti-epileptic drugs can make him more sensitive to the effects of alcohol, and that alcohol can decrease the efficacy of his medication
- ○ Advise him to be aware of his own seizure triggers, eg lighting/late nights/missed meals
- ○ Tell him that having epilepsy may influence his career choice (eg the armed forces won't consider people with epilepsy)
- ○ www.epilepsyresearch.org.uk is a good source of patient-orientated information about the effects of epilepsy on lifestyle.

20. An 8-year-old girl has had three afebrile seizures. You decide to start treatment. Talk to her parents about starting anti-epileptic medication

- ○ Explain the reason for starting medication (preventing further seizures)
- ○ Explain how medication is administered (gradually increasing dose: start low/go slow)
- ○ List the main side-effects and what to do if the parents notice them – don't stop medication (needs to be tapered off), and contact the epilepsy team urgently
- ○ Explain when to review medication – no obvious benefit/unacceptable side-effects/at puberty/seizure free for 2 years
- ○ Main side-effects of:
  - Sodium valproate:
    - Gastric upset
    - Hair loss (transient: regrowth after 6 months)
    - Sedation
    - Change in behaviour (hyperactive/aggressive)
    - Teratogenicity
    - Hepatotoxicity (some clinicians check liver function tests regularly)
  - Carbamazepine:
    - Urticaria
    - Nausea/ataxia/visual changes (respond to more gradual dose increments)
    - Bone marrow suppression (need to tell parents what to look out for)
  - Lamotrigine:
    - Rash
  - Topiramate:
    - Headache/tiredness/weight loss
    - Memory/concentration difficulties
    - Nephrolithiasis.

21. Explain to this medical student the various different insulin regimes for children with diabetes

- Use diagrams/graphs to illustrate twice-a-day injections, basal bolus regime, insulin pump therapy
- Talk about advantages and drawbacks with each type:
  - Basal bolus:
    - Appears more physiological
    - More flexibility, greater control over what and when to eat
    - More injections (at least four a day)
    - Able to 'carbohydrate count' – adjust insulin requirements to meal characteristics
    - Need to inject at school (can be a problem for children and for schools)
  - Twice a day (used rarely and new patients will not be started on this regime):
    - Fewer injections
    - Need for regulated snacks to prevent hypos
    - Timing of meals important
  - Insulin pump:
    - Precise control of insulin dose
    - Need for frequent testing
    - Need to be carbohydrate counting
    - Attached 24 h per day.

22. This 1-year-old child has been diagnosed with an egg allergy. Talk to her mother

- The more processed and cooked the egg, the less allergenic it will be: talk about which forms to avoid
- Explain that there is a strong likelihood that she will grow out of it
- Consider an egg challenge after her second birthday depending on skin prick tests/specific IgE results. Some challenges are left until later if tests still very suggestive of ongoing allergy. Co-existent allergy/other medical problems may be relevant to the timing
- Arrange dietician input.

23. You have prescribed an EpiPen® for this child with a peanut allergy. Talk to her parents

- Explain that anaphylaxis is life-threatening
- List the symptoms to watch out for (breathing problems, swelling in lips or mouth, pale, vomiting, drowsiness)
- Explain how to inject
- Pens for home and school
- Second pen needs to be available as some children need extra pre-hospital dose of adrenaline

- ○ Arrange training for everyone who looks after child (including school) – allergy specialist nurse will help with this
- ○ Don't let pens go out of date
- ○ Recommend a training video (check one is available in your department).

24. Talk to the mother who has a jaundiced neonate and who is unhappy that she is not able to take her baby home (because the baby needs phototherapy)

- ○ Explain the cause of jaundice (breakdown of red cells, immature liver)
- ○ Explain the risks of jaundice – bilirubin crosses into the brain, causes drowsiness/poor feeding. Very high levels in children who are already sick can be more serious; permanent damage can be caused
- ○ Treatment: exchange transfusion – avoid need for this by phototherapy/adequate hydration
- ○ Draw diagram of a bilirubin risk chart
- ○ Explain the need for blood tests and the likely length of stay in hospital.

25. Talk to this mother about the implications of premature delivery at 23 weeks

- ○ Consider the following before talking to Mum:
  - Mum's support network
  - How likely she will retain information (is she in active labour?)
  - Her previous experience of preterm deliveries
  - Any known antenatal problems so far
- ○ 'The most important thing to tell you is that this is a very high-risk situation and by that I mean your baby may not survive'
- ○ Many do not survive delivery (roughly half)
- ○ Of those admitted to the newborn unit many do not survive (look at Epicure 2 data www. epicure.ac.uk)
- ○ Explain that the baby will be on the neonatal unit for 4–5 months, and that all baby's 'systems' are immature and will need support; the main problems are with breathing, feeding, infection and bleeding into the brain
- ○ Explain that many have long-term problems with development; particularly movement, vision and hearing problems
- ○ Outline what will happen immediately after the baby is born (neonatal team present, intubation, transfer to NNU).

26. This child has recently been admitted with a painful sickle crisis. Talk to her mother about pain relief

- ○ Stepwise (paracetamol, ibuprofen, opioids)

- Explain that they should come to hospital if the pain is severe (requiring opioids); also, explain the need to come to hospital for a high fever, pain in the chest, spine or abdomen, neurological signs
- Don't forget other aspects of management of a crisis (think about rest, hyper-hydration, oxygen, antibiotics).

27. **Talk to the mother of this child with diabetes on multiple daily injections about the management of hypoglycaemia**
    - When to treat low sugars ('4 is the floor')
    - Symptoms – may be none, pallor/shaking/sweating followed by irritability/change in behaviour/drowsiness
    - How to treat mild symptoms (dextrose tablets or 50 ml Coke/Lucozade followed by long-acting carbohydrate, eg biscuits/bread/banana)
    - Hypostop® if drowsy, glucagon if unconscious
    - Explain that there is an increased risk associated with recurrent hypoglycaemia, alcohol, lots of exercise
    - Check understanding, offer written information and a diabetic nurse follow-up, who will go through this information again and have more information on how to treat.

28. **The mother of a 4-month-old baby admitted with bronchiolitis requiring nasal cannula oxygen and nasogastric feeds is demanding antibiotics for her son. Explain what bronchiolitis is and the management.**
    - Establish knowledge
    - Explain the difference between viruses and bacteria, and what antibiotics can and cannot treat
    - Explain 'supportive treatment'
    - Reassure her that you will regularly be reassessing her baby.

29. **Talk to the mother of this 12-week-old baby, who has gastro-oesophageal reflux but has good weight gain, who is wanting a prescription of 'something to treat the discomfort and vomiting'**
    - Explain that it is a very common problem for babies and that 'almost half of all babies will experience reflux'
    - Talk about optimising feeding – keeping upright after feeds; however positional changes in sleeping babies are not recommended
    - Depending on how severe the reflux is there are treatment options such as feed thickeners and alginate therapy
    - Reassure her that the vast majority will improve with time and completely resolve within the first year of life while empathising that things are difficult now
    - Arrange to see her again in a few weeks to see how they are getting on
    - There are NICE guidelines on the diagnosis and management of gastro-oesophageal reflux at www.nice.org.uk/guidance/ng1.

30. **Talk to this 15-year-old girl with anorexia with postural hypotension and significant weight loss in the last 2 weeks about the decision to admit her into hospital**
    - ○ Establish common ground – what does she understand so far?
    - ○ Outline current concerns – life-threatening condition, her body is not able to maintain blood pressure, ie it is not coping with this weight loss, low blood pressure can make you feel really unwell/dizzy/make you faint
    - ○ Plan for admission – bed rest, adequate fluids and plan for portion size (ie start with 1/4 portions)
    - ○ Explain other members of the multidisciplinary team (dietician, psychiatrist, paediatrician) will be involved and come and see her
    - ○ Explain she will need daily blood tests (looking for re-feeding syndrome).

31. **A 10-month-old boy has been diagnosed with developmental hip dysplasia. Talk to his angry parents**
    - ○ Spend time listening
    - ○ Explain that every effort is made to pick up abnormal hips clinically but the condition is not always able to be diagnosed at birth
    - ○ Explain that some cases are missed, and that of these many resolve spontaneously
    - ○ Explain that we do not offer ultrasound screening to everybody.

32. **Talk to the parents of an 18-month-old child who has been seen several times with uncomplicated viral infections. Her parents are demanding tests of her immune function**
    - ○ Explain that immune function is investigated for recurrent severe, prolonged, or unusual infections
    - ○ Explain that repeated viral infections are normal for this age group (at least eight per year is normal)
    - ○ Explain that there is some evidence to suggest that viral infections in preschool children can have a beneficial effect on the immune system (reducing allergy)
    - ○ Normal growth is reassuring
    - ○ Offer follow-up.

33. **Give advice to this mother who is worried because her 2-year-old son is a very fussy eater**
    - ○ Most toddlers have 'good' and 'bad' days when it comes to eating
    - ○ Most toddlers are capable of regulating their own intake of food and drink. It is very reassuring that this boy is growing well, as this means that he is probably eating enough

- ○ Encourage the family to eat together, away from distractions such as television
- ○ Explain how to recognise cues that the child has had enough (eg saying 'no', keeping their mouth closed when food is offered, pushing away the spoon or bowl, holding food in the mouth and refusing to swallow it), and when to stop offering food if the child is refusing it
- ○ Check that the child is not consuming too much milk or snacks in between meals
- ○ Discourage drinking large amounts of juice or squash, especially between meals
- ○ Encourage the use of finger foods rather than parent-led spoon feeding. Allow the toddler to take control of his intake, to handle the food and to make a mess with it. Give him many small portions of different foods
- ○ Avoid using puddings as a reward for eating (as this discourages the child from enjoying the first food that is offered)
- ○ Provide written advice and health visitor follow-up to give the family support and monitor the child's growth.

34. This mother is worried about her 5-year-old son who still wets the bed at night. Consultation has excluded underlying medical problems. Talk to her about the non-medical management of enuresis

- ○ Explain that around 15% of 5-year-olds have night-time incontinence (bed wetting)
- ○ Explain that the child is not wetting the bed on purpose, and has no control over whether or not he is incontinent at night
- ○ Avoid giving him blackcurrant juice/squash (which irritate the bladder) and any drinks containing caffeine
- ○ Only give him water to drink after his evening meal
- ○ Treat constipation if there is any history of it
- ○ Make sure that it is easy for him to get to the toilet overnight (consider using a nightlight)
- ○ Encourage him to drink plenty of fluid during the day (aim to get in full daily intake by mid-afternoon, which in this age group is 1–1.4 litres)
- ○ Encourage him to hold on, to increase bladder capacity
- ○ Make sure that his bladder is empty before he goes to sleep
- ○ Give behavioural rewards for those aspects of good drinking and toileting behaviour over which the child has gained control (eg drinking plenty of fluid during the day, going to the toilet before bedtime)
- ○ NICE guidelines on the management of nocturnal enuresis were published in 2010 (www.nice.org.uk/cg111).

35. **This mother explains that her 6-year-old daughter who has primary nocturnal enuresis would like to go on sleepovers. Talk to the mother and explain the use of desmopressin**
    - ○ Describe how desmopressin works
    - ○ Explain how it should be taken, last thing at night, with nothing to drink other than water to quench the child's thirst until she has passed urine in the morning
    - ○ Start at a lower dose (refer to latest *BNF for Children* for dosing information) and increase to the higher dose if this is not effective
    - ○ Discuss the side-effects of desmopressin (water overload) and how to avoid them
    - ○ Do a 'practice run' at home. Desmopressin can be used for sleepovers once the effective dose is known
    - ○ Explain that you will provide written advice as part of your summing up.

36. **This 3-year-old girl has been diagnosed with iron-deficiency anaemia. Talk to her mother about ways to increase the amount of iron that she obtains from her diet**
    - ○ Discuss the effects of iron in the body (it is required to make haemoglobin, which carries oxygen around the body)
    - ○ Describe the symptoms of iron deficiency (malaise, reduced appetite, decreased concentration and learning)
    - ○ An iron-rich diet contains at least one source of iron per day
    - ○ Good sources include beef, turkey, most oily fish, breakfast cereals enriched with iron (read the label) and green vegetables such as broccoli and spinach
    - ○ High-fibre foods and vitamin C increase iron absorption
    - ○ Limit the intake of cow's milk
    - ○ Avoid tea and other caffeine-containing drinks, particularly around mealtimes
    - ○ Asking 'Are there any of these foods that your child particularly enjoys?' is a good way to check the mother's understanding
    - ○ Offer to provide written information about iron-rich foods for children.

37. **This 9-year-old boy has had an intermittent facial twitch**
    - ○ Talk to his parents about motor tics
    - ○ Check whether the parents have heard this term before and what they understand by it
    - ○ Explain that transient tic disorders are common among school-age children
    - ○ Describe some common motor tics (eg eye blinking, sticking out the tongue, shrugging the shoulders, grimacing, coughing, lip biting)

- Rarely tics can be associated with infections such as streptococcal infection/Lyme disease or some viruses such as EBV. These will be tested for and treated
- Tics are often exacerbated by stress, heat, being tired or excited, or becoming self-conscious about the tic
- Remember that tics are involuntary, and children are unlikely to be able to have control over them all of the time, although occasionally they can suppress the tics for short periods
- Ideally, class peers should be educated about tics, and encouraged not to draw attention to them. It can be helpful to suggest to the child a way that they can explain about the tics to other children who ask about them
- Offer to provide written information for the child's school and for other family members
- Tell the mother to seek help if the tics become more frequent or severe, or become associated with other worrying behaviour (eg aggression, depression or obsessive compulsive signs), or are causing emotional problems or interfering with everyday life
- The only proven beneficial treatment is cognitive-behavioural therapy which can be arranged if tics are persistent/progressive/intrusive. (European clinical guidelines for the management of tic disorders can be found on bestpractice.bmj.com)

38. Talk to this mother about the MMR vaccine; she is concerned about the links with autism and inflammatory bowel disease, as well as exposing her 1 year old to 'too much in one go'

- Establish what Mum's concerns are and from what sources
- Empathise, 'It can be confusing with so much conflicting information, I understand you are just trying to do what's best for your child'
- Talk through each concern – very safe vaccine which has been extensively studied
- Talk about the research that suggested a link between autism and inflammatory bowel disease, how this has been discredited and how it has not been reproduced in any study before or since
- Avoid being pushy or judgemental. It is the parent's responsibility to weigh up the risks and benefits, and the paediatrician's responsibility to provide accurate, easily understandable information to aid that choice, while acting in the child's best interests
- Don't forget to discuss the risks of the diseases being vaccinated against
- Further information can be found on the Department of Health website (www.gov.uk). 'The Green Book' is an excellent resource and discusses the risks and benefits of all vaccines.

39. You are giving some sedation to a child for a CT scan

- ❍ Check the parents' current understanding and acknowledge their concerns
- ❍ Explain the reasons why sedation is needed (ie explain the intended benefits):
  - To avoid causing unnecessary distress
  - To maximise the likelihood that the procedure will be successful
  - Because the procedure is important – describe the difference that it will make
- ❍ Explain how the sedation will be administered:
  - By mouth/intravenously/intranasally
- ❍ Discuss the potential side-effects, and what will be done to minimise this risk:
  - Risk of respiratory depression – oxygen will be available, a doctor will be supporting the airway and monitoring the child's oxygen levels, and an antagonist (eg flumazenil) will be available
  - Risk of aspiration – the child will be kept nil by mouth
  - Risk of failure to sedate
- ❍ Make sure that the parents have understood what you have told them, ask whether they have any further questions, and ascertain whether or not they are happy to give consent.

40. This mother is refusing to give her newborn baby vitamin K. Talk to her about this

- ❍ Find out what the mother already knows about vitamin K and what her concerns are
- ❍ Explain why vitamin K is recommended
- ❍ Administering vitamin K reduces the risk of haemolytic disease of the newborn, which is a serious bleeding disorder:
  - Haemolytic disease of the newborn (HDN) may have an early onset (within 2–4 hours), which is very rare, or may have classic onset (1–2 weeks) or late onset (4–6 weeks), which occur in 4–8 per 100 000 births
  - Risk factors include breastfeeding (which results in slower build-up of levels of vitamin K), liver disease, traumatic delivery (where there has been bruising or bleeding), prematurity and the use of maternal anticonvulsants in pregnancy
  - HDN is very rare compared with childhood cancers. However, HDN is considered to be completely preventable if vitamin K is administered, whereas the increased risk of childhood cancers is unproven (see below)
- ❍ Vitamin K is not found in many foods, and breast milk contains only small amounts. It is normally produced by 'helpful bacteria' in the gut. It takes time for babies to build up their own stores of vitamin K

- There was controversy about the safety of intramuscular vitamin K after a case-control study was published in 1990 that looked at potential causes of childhood cancers and found an increased risk of cancer in children who had received intramuscular vitamin K. However, many subsequent studies have found no evidence for any such association
- The following problems are associated with the use of oral vitamin K:
  - Concern about poor compliance
  - The optimum regimen is not known
  - Some studies have shown it to be less effective in preventing HDN than vitamin K administered intramuscularly
- The normal oral regimen is one dose at birth, one dose on day 4–7 (with the Guthrie test) and one dose at 4 weeks for breastfed babies
- The decision as to whether to use vitamin K is ultimately that of the parents
- Offer to provide written information.

PART III

Clinical stations

7

INTRODUCTION AND SUGGESTED APPROACH

The clinical stations are seen by many candidates as the most challenging part of the examination. However, thorough preparation and honing of clinical examination technique will give you more confidence – even if you encounter a case you may not have seen before.

7.1 Preparation and technique

Preparation and technique are intertwined. Many candidates fail the exam because of poor 'short case' technique, and this is usually down to poor preparation of potential exam scenarios and a lack of awareness of what is expected of them in each station. The well-prepared candidate will give themselves many opportunities to pick up marks, just as the poorly prepared candidate will inevitably drop marks.

In particular, if your examination technique is poor you will fail.

The RCPCH Anchor Statements for 'Expected Standard/Clear Pass' demonstrate that three key areas are assessed:

- **Conduct of examination** – Introduces oneself, puts parent and child at ease. Displays an appropriate level of confidence. Appropriate pace without rushing. Acknowledges child fully and explains intended examination if deemed appropriate. Adjusts language and behaviour to suit age of child.
- **Clinical examination** – Systematic and uncluttered technique. Able to identify clinical signs and interpret their meaning.
- **Discussion with examiners** – Sensible differential diagnosis. Able to suggest a sensible management plan, including investigations. Demonstrates an understanding of impact of findings on patient and family unit.

For example, if you are asked to examine the respiratory system, to pass the exam you will need to do more than just pick up and present findings. It may be you will need to present not only the most likely diagnosis, eg cystic fibrosis, but also suggest the different investigations that could have led to this diagnosis, such as the sweat test, genetic analysis or the Guthrie test looking at immunoreactive trypsin. This reiterates the philosophy that the exam reflects everyday clinical practice.

- Examine the child
- Pick up and present the abnormalities
- Answer questions relevant to the case

It is essential to be completely clear about how to examine any system. Decide on your own method, learn it and practise it. Use this method throughout the preparation period when seeing short cases. Try not to change your approach in the run-up to the exam. Remember there is no definitive way to examine any system; you need to find your own scheme. Whichever scheme you use, remember that the examiner is unlikely to tolerate imprecise or clumsy clinical examination, and this should be avoidable with proper preparation. This book suggests an examination scheme for most potential scenarios. You are also referred to standard texts of paediatric examination, and up-to-date information regarding the examination from the Royal College of Paediatrics and Child Health website (www.rcpch.ac.uk).

Remember that confidence in the exam comes from knowing that your examination technique is not only correct but is also **reproducible in a highly stressful environment**. This comes from relentless practice of examination and presentation in the company of several different 'examiners'. It is important to be mock-examined by a range of colleagues, some of whom you know and some of whom you don't. This reflects the six different examiners who will soon be assessing you in the clinical stations with no prior knowledge of your performance in a previous station, or indeed your performance in the workplace. At the very least you should go around in pairs and be very objective when assessing each other's performances. It will help to take a copy of the Anchor Statements with you to see how you measure up to the Expected Standard.

During the examination itself there are several common pitfalls:

- Poor engagement with the child, including failure to look at a child to judge whether you may be eliciting pain. For example, repeated examination of an enlarged spleen may start to become uncomfortable for a patient if you are the fourth consecutive candidate to have seen them. It is important to be empathetic with the patient, just as you would be on the wards.
- Failure to talk your way through an examination when it has been requested/is appropriate.
- Lack of conviction in your findings, enabling an examiner to place doubt in your mind and throw you off balance. For example, a candidate was asked to examine an abdomen with bilateral masses and then asked to present her findings. She presented the patient as having an enlarged spleen and liver. In the subsequent discussion the examiner was able to change the candidate's mind twice as to whether the masses were bilaterally enlarged kidneys or a large liver and spleen.
- Stopping the examination when you have identified the organ in question. For example, there is a set routine for examining an enlarged liver and you must continue with your examination until you are stopped. Another candidate was asked to examine the abdomen. There was an enlarged liver and the child had a fever, having just returned from Africa. The candidate correctly identified malaria as the diagnosis but failed the short case. On counselling she was told that her examination of the liver was inadequate because she did not percuss its size.
- Excluding a clear physical sign if you feel it does not fit in with other signs. Another candidate was asked to examine the cardiovascular system and to comment afterwards. She correctly identified the missing left brachial pulse and the left thoracotomy scar, making the correct diagnosis of repaired aortic coarctation. However, this candidate also heard a soft ejection systolic murmur and could not fit it into the picture and did not mention it. Of course, such a murmur is to be expected in coarctation. Despite this the candidate passed.
- Failing to come up with some common differential diagnoses in the discussion. The differential diagnoses in this text are 'practical differential diagnoses' focusing primarily on common scenarios and ones that are likely to be seen in the exam. Many candidates report that they are not given the chance to present a full differential diagnosis, but are asked for one or two possibilities relating to the child they have seen. Where relevant, the most likely differentials will be at the top of the list in this book.

Basic format for clinical examination

- ○ Introduce yourself to the child and parent
- ○ Be prepared to talk about your findings as you proceed
- ○ Observe and be prepared to comment on:
  - General appearance and health – well or unwell? Ward patients are occasionally used
  - Dysmorphic features – syndromic or otherwise
  - Growth and nutrition – note obvious abnormalities. You may be asked to assess pubertal status
  - Development – neurodevelopmental abnormalities may be noted during examination of other systems. Many patients are eligible for two or more stations per exam session, for example neurology and child development
  - Hands – colour, clubbing, nail abnormalities, poor perfusion
  - Face – cyanosis, anaemia, jaundice
  - General observations such as presence of a nasogastric tube
- ○ Palpate
- ○ Auscultate
- ○ Ask for permission to examine any other relevant part of the body
- ○ Describe your findings
- ○ Answer questions about your findings until the bell rings for the end of the station

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7.2 Presentation skills

Presentation skills are important. In particular, the first few sentences you say about a certain scenario need to be practised. Examples of how you should present cases are included in Chapters 8–13. When presenting, it is useful to have a few general words and phrases that can apply to most cases, eg 'Harry is a boy who looks well grown and I would like to plot his weight on a centile chart ...'. This approach overcomes nerves and gives you time to compose yourself.

The information in this section of the book is presented partly in note form and partly as it would appear in the exam, using case scenarios to illustrate the knowledge required on a particular topic. Additional information pertaining to many of the conditions seen in this section can be found in Parts I and II.

8

CARDIOVASCULAR

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8.2 Clinical examination
    Suggested approach to cardiovascular examination

8.3 Cases
    Case one: Complex cyanotic congenital heart disease
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    Case three: Cavo-pulmonary shunt
    Case four: Innocent murmur
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    Innocent murmurs
    Dextrocardia
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    Coarctation of the aorta
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    Fallot's tetralogy
    Eisenmenger syndrome

Cardiomyopathy
Infective endocarditis
Syndromes associated with cardiac lesions
Surgery for congenital heart disease
Chest X-ray findings

8.1 Introduction

Preparation

The cardiology station can be a hard part of the exam. In this chapter we have identified some of the important aspects of the examination and more common conditions seen. Bear in mind advances in cardiac surgery have meant that there are many children who have had complex congenital heart disease, and it may be unrealistic to be able to diagnose their exact pathology on examination; you should however be able to discuss your clinical findings and make suggestions as to what could be the cause of them. It is essential to have a good system of cardiovascular examination that you are 'fluent' in and some background knowledge (read through paediatric cardiology in one of the standard texts) to help with the interpretation of physical signs. The best places to see children with signs are on the cardiology ward (pre-elective surgery) and in the cardiology clinic. These children will be most representative of the cases you are likely to see in your exam. You should ask a cardiology specialist trainee or consultant to critique your examination technique and point out to you how they would best elicit the relevant signs. There are many important physical signs that you may never have seen outside a specialist clinic/ward. They are often surprisingly easy to elicit and once seen they are often not forgotten.

During the examination

It is important to be able to adapt the routine of your examination by being responsive to the child and what they will tolerate, so that you have the best chance of eliciting the clinical signs. It is not unreasonable in the case of a small child to move straight on to auscultation at the very beginning if the patient is quiet, to listen to heart sounds and murmurs before the child becomes upset. It is probably best to explain yourself to the examiner if you perform the examination this way.

However another dilemma is what to do if the examiner asks you to 'examine this child's heart'. Do you just auscultate the heart? Or carry out the full cardiovascular examination? Clearly, if the examiner asks you to perform a cardiovascular examination then the full examination should be done. If asked just to examine the heart, in our experience it is usually best to do the full cardiovascular examination; the examiner can then be more specific and tell you to take shortcuts if that is what they are looking for. If you are unclear what is expected of you, then it is perfectly reasonable to ask the examiner directly.

Taking blood pressure and checking jugular venous pressure (JVP; see Section 8.4) are part of a complete examination and you should include them in all your practice cases so that you get used to doing them quickly and under pressure for children at a variety of ages. Because they can often take up a disproportionate amount of time and are so often normal, it may be acceptable to leave them to the end, but you should make every effort to let the examiner know that you haven't overlooked them and give the examiner the prerogative to stop you from attempting them if he or she does not think they are relevant. If in doubt – do it!

Particularly with the cardiovascular system, there are many signs that have the potential to give very accurate diagnostic information, but require years of experience to be fully appreciated, such as split-second heart sounds and abnormal jugular venous pressure (JVP) waveforms. As a general rule, if a sign is not obvious it is probably not there or not worth mentioning. Don't spend so long on these aspects of the examination that you neglect the bigger picture.

In most short case scenarios it is sensible to talk your way through the examination of the case. In the cardiovascular system examination however, particularly if the diagnosis is not obvious, it is wise to keep your own counsel. Clearly if you see something obvious, eg Down syndrome, cyanosis or chest wall scarring, it is appropriate to mention it. Many of the signs you discover will be much easier to interpret once you have gathered all the information, particularly when it comes to interpreting murmurs. You would be wise to not volunteer any information about heart sounds and murmurs until you have listened as fully as you intend to, including checking for radiation and response to manoeuvres. This will give you the maximum time to assimilate all the information and put it together into a cohesive, reasonable-sounding interpretation.

The key to success in this station is to be able to confidently carry out a thorough examination, to be able to detect the relevant signs and to be able to formulate them into a sensible differential diagnosis. Furthermore you must have enough background knowledge of the more common conditions to be able to answer some questions about the significance of your findings and further management.

8.2 Clinical examination

Suggested approach to cardiovascular examination

Examine this child's heart.

This boy had a cardiac procedure as an infant, please examine his cardiovascular system.

Initial procedure

- Introduce yourself to the parents and the child.

General observations

- Age
- Growth and nutritional status: growth is crucial to determining management in congenital heart disease, hence is particularly important in this station
- Colour: pink, pale or blue (or jaundice as in Alagille's)
- Dysmorphic features (could give clue to underlying cardiac lesion)
- Monitoring (may have oxygen saturation probe)
- Supplemental oxygen
- Increased work of breathing (recession, tachypnoea)

Hands to arms

- Perfusion: cool or warm hands, peripheral capillary refill time, should be less than 2 s
- Clubbing: if you are unsure whether it is present, don't spend a long time looking for it, as it may not be significant enough to comment on. Best to look for 'loss of diamond' when comparing index fingers (first sign of clubbing). Clubbing is present after long-standing arterial desaturation (over 6 months). Could be present in pink, well-saturated children if they have recently had corrective cardiac surgery (or if it has a 'non-cardiac' cause)
- Fingernails for signs of infective endocarditis: see notes below
- Radial pulse and brachial pulse: compare both sides:
  - Make a show of counting pulse rate (ie with a watch/clock, count for 10 s x 6)
  - Comment on rate rhythm, character (you should know normal rate parameters for different age groups; see Box)

Hyperconvex/hypoplastic nails	Turner syndrome
Short 4th and 5th metacarpals	Turner syndrome
Single palmar crease	Down syndrome
Long tapering fingers	DiGeorge syndrome
Arachnodactyly	Marfan syndrome
Radial thumb abnormalities	VACTERL, Holt–Oram syndrome
Overlapping fingers	Trisomy 18
Polydactyly	Trisomy 13

Table 8.1: Syndromes with hand abnormalities and heart defects

Normal ranges for heart rate (resting) in beats per minute

Neonates	110–150
2 years	85–125
4 years	75–115
6 years and over	60–100

- Brachial/carotid pulse characters:
    - *Collapsing*: run-off lesions, classically aortic regurgitation, could also be seen in patent ductus arteriosus, large arteriovenous fistula
    - *Thready*: congestive cardiac failure, circulatory shock
    - *Asymmetrical*: post-coarctation repair (using L subclavian flap)
    - *Slow rising:* flattened pulse amplitude, implies impaired ejection from left ventricle (eg aortic stenosis)
    - *Pulsus paradoxus*: cardiac tamponade/pericarditis, severe asthma/high intrathoracic pressures

Tip +

Don't suggest the pulse character is abnormal unless it is very obviously abnormal. If you are unsure, it is less likely to be significant and you should move on

Blood pressure

- You should make a judgement about how quick and easy it would be to check the BP manually at this point. In older children this can be done rapidly and you should do it. In younger, less co-operative children it can take a while to do this but it is still an essential part of a complete, structured examination. If you plan to come back to BP at the end then make sure the examiners are aware of this or it may appear that you have simply forgotten. You will need to practise doing this on children of various ages so that you don't take too long over it in the exam. Your approach should be:
    - Explanation
    - Position child as you want them to be
    - Select correct cuff size
    - Inflate the cuff while palpating the brachial/radial pulse until the pulse disappears
    - Inflate the cuff a further 10 mmHg
    - Place the stethoscope over the brachial artery
    - Deflate the cuff slowly until regular sounds (the Korotkov's sounds) can just be heard through the stethoscope
    - Note the systolic pressure as accurately as possible
    - Continue to deflate the cuff slowly; the sounds get louder then suddenly become muffled and disappear
    - Record the point at which the sounds become muffled as the diastolic pressure
    - Interpret the results
- All children need an explanation of what is about to happen. This should start with *'have you had your blood pressure checked before?'* Young children need very careful reassurance and this should be well practised. It is likely that if the examiner has indicated that you should take the blood pressure of a young child then there will be a lot riding on your sensitive, paediatric approach
- Make sure you choose the correct cuff – cuffs that are too small may cause a falsely high BP. Cuff widths should be approximately two-thirds of the length of the upper arm (or 50% of the circumference of the arm), and the bladder should encircle the limb almost completely
- Technically you should take at least two readings before you say what the blood pressure is. This is probably only necessary if you have been specifically asked to check the BP. You should let the examiner know what the result was from the first reading and then say *'I would normally confirm this with a second blood pressure reading'*
- It is extremely unlikely you will get a child with a coarctation that is preoperative, however you may be asked about what you might find on checking their blood pressure (ie a significant difference between the right and left arm blood pressure – pre- and post-ductal)
- *In reality BP is often measured by a Doppler machine or Dinamap®*. If you are not familiar with how to use these you should ask one of the nursing staff to teach you. Don't be caught out by not being able to use a simple piece of equipment like a BP monitoring machine!
- Interpreting whether BP readings are normal is difficult. There are published normal ranges based on the child's age and height. As a rough guide a systolic BP should be

no more than 90 plus double the child's age. If you find a BP that is outside this range you should check a second reading and comment on the fact that it is not a normal reading but that you would check the value against a table of normal ranges. Table 8.2 is a rough guide.

Newborn	Mean BP should be equal to or above gestational age (eg mean of > 40 mmHg for term infant)
Infancy	80–95 mmHg systolic
Preschool	80–100 mmHg systolic
School age	90–110 mmHg systolic
> 12	100–120 mmHg systolic

Table 8.2: Approximate values for blood pressure in children

Face and neck

- Conjunctival pallor (approach with care)
- Sclera – jaundiced/injected
- Central cyanosis – becomes clinically apparent when saturations are below 85%. This is not necessarily true if the patient is anaemic or dark skinned. You should say that the patient is not clinically cyanosed unless they are very obviously blue. In practice, for the purposes of the exam, if you are not sure and the patient is not clubbed, do not spend a long time looking
- Mouth – again do not spend a long time looking, but if the patient phenotypically is Marfanoid, you can look for a high arched palate. Look for dental caries
- Carotid pulse character
- Jugular venous pressure – in older children (see notes below)

Chest

Inspection

- Expose fully wherever possible
- Shape (chest wall deformities)
- Comment on any visible impulses (could indicate hyperdynamic apex beat)
- Scars – ask the child or parent to point out any scars on their chest (see Box)

Causes of chest wall scars

Right lateral thoracotomy

○ Modified BT shunt
○ Lung causes (lobectomy)
○ Tracheo-oesophageal fistula repair

Left lateral thoracotomy

○ Modified BT shunt
○ Coarctation repair
○ Patent ductus arteriosus ligation
○ Pulmonary artery banding
○ Lung causes (lobectomy)

Median sternotomy

○ Any bypass surgery (most corrective surgery of intracardiac abnormalities requires bypass)

Chest drain scars

Mediastinal drain scars

Chest wall pacemakers or reveal devices (often in left pectoral region)

Palpation

- Apex beat (right or left): the first and most important thing is to look for dextrocardia at this point; if you miss it now it will be difficult not to look foolish when you eventually realise. Use both hands, place a hand on each side of the chest and remove the hand that hasn't got the strongest impulse underneath it (if you detect dextrocardia you must remember to look for associated abnormalities/ Kartagener syndrome)
- You can then proceed to determine:
  - Site: normal (4th or 5th intercostal space, mid clavicular line) or displaced
  - Character: hyperactive characterises lesions with volume overload (large left-to-right shunt/valvular regurgitation)
  - Right ventricular heave: a palpable right ventricular impulse indicative of high right-sided pressures (for example, right ventricular outflow tract obstruction)
- Thrills:
  - Upper left sternal border – pulmonary stenosis (rarely patent ductus arteriosus)
  - Upper right sternal border – aortic stenosis
  - Lower left sternal border – ventricular septal defect
  - Suprasternal notch – aortic stenosis (occasionally pulmonary stenosis, patent ductus arteriosus, coarctation of the aorta)
  - Carotid arteries – aortic stenosis (occasionally coarctation of the aorta)

Auscultation

Listen in four areas:

1. Mitral (apex)
2. Tricuspid (lower left sternal edge)
3. Aortic (right 2nd intercostal space)
4. Pulmonary (left 2nd intercostal space)

After auscultating the heart you can quickly listen to the base of the lungs for inspiratory crackles (pulmonary oedema secondary to congestive heart failure).

Several candidates have been asked to point out the areas to the examiner.

Remember this is on the assumption that the heart is structurally normal and dextrocardia is not present.

Heart sounds

Take note of these before assessing the nature of any murmur:

- Are they both present?
- First heart sound – loud when cardiac output high, eg when nervous or excited, anaemia and pyrexia. Soft when cardiac output is low or there is poor left ventricular function
- Is the second heart sound normal?
- If the second heart sound is louder than the first when listening over the apex, then this could indicate pulmonary hypertension

The following are classically heard by consultant cardiologists and medical students but very rarely by the rest of us. Once again, if it is not obvious, don't spend a long time looking for it, as it is unlikely to be significant for the purposes of the exam. You should however know the significance and implications of the following, and how to detect them if they are there:

1. Splitting of second heart sound:
    - Pulmonary valve closes later than aortic valve in the normal cardiac cycle
    - Best listened for at the upper left sternal border
    - Increased separation between sounds in inspiration and decreased separation (becoming single second heart sound) in expiration are normal
    - Widely split: classically occurs in atrial septal defect (where the right ventricle is overfilled and pulmonary valve closure is delayed), mild pulmonary stenosis and right bundle branch block
    - Fixed splitting of the second sound (atrial septal defect): when there is no variation with respiration, the two atria are functioning as one and respiration has the same effect on both the pulmonary and systemic circulations
    - Single S2 occurs when there is pulmonary hypertension (pulmonary valve closes quickly), transposition of the great arteries, or abnormal aortic or pulmonary valves (atresia or severe stenosis)

- Paradoxical splitting (aortic valve closes last) – left bundle branch block, severe aortic stenosis

2. Third heart sound ('Tennessee'):
   - Often heard in normal children
   - Can indicate reduced ventricular compliance
   - Best heard with bell over apex/lower left sternal border
3. Fourth heart sound ('Kentucky'):
   - This occurs shortly before the first heart sound
   - Almost always pathological but rare in children
   - Indicates reduced ventricular compliance
4. Gallop rhythm:
   - Implies loud S3 or S4 and tachycardia
   - Always pathological
5. Ejection clicks:
   - Classically occur in aortic valve and pulmonary valve stenosis, but may also be heard when there is a large dilated aorta in tetralogy of Fallot, coarctation of the aorta, patent ductus arteriosus or when there is a dilated pulmonary artery in pulmonary hypertension. The sound is made by the forceful opening of the valve
   - Aortic click – lower left sternal edge and apex
   - Pulmonary click – 2nd left intercostal space

Murmurs

Make sure you have fully assessed a murmur before you comment on it. A full assessment consists of the following six components:

1. Timing in relation to cardiac cycle: patterns of murmur are given in Table 8.3

Systolic	*Diastolic*	*Continuous, ie overlaps both heart sounds, therefore is not confined to systole or diastole*
Ejection systolic (crescendo/decrescendo) • Could be innocent • Aortic stenosis • Pulmonary stenosis Pansystolic • VSD • Mitral regurgitation • Tricuspid regurgitation Systolic click (occurs mid-systole) with late systolic murmur • Mitral valve prolapse Ejection click (occurs early in systole) • Aortic valve disease	Early diastolic murmur • Aortic regurgitation (best heard at LLSB, with bell, in expiration, sitting forward) Mid-diastolic • Mitral/tricuspid stenosis Opening snap/diastolic rumble • Mitral stenosis	• PDA • ASD • Blalock–Taussig shunt • AV malformation • Aneurysm • Collateral vessels • Venous hum • Peripheral pulmonary stenosis • Aortopulmonary window

Table 8.3: Patterns of murmur. ASD, atrial septal defect; AV, arteriovenous; LLSB, lower left sternal border; PDA, patent ductus arteriosus; VSD, ventricular septal defect

2. Site of maximal intensity (see Table 8.4)

Remember this is on the assumption that the heart is structurally normal and dextrocardia is not present.

Upper left sternal edge = pulmonary area	*Upper right sternal edge = aortic area*	*Lower left sternal edge*	*Apex*
• Pulmonary stenosis • Pulmonary flow murmur • Pulmonary artery stenosis • PDA • RVOT obstruction (eg Fallot's)	• Aortic valve stenosis • Subaortic stenosis • Supravalvular aortic stenosis	• Aortic regurgitation (loudest over 3rd or 4th intercostal space) • VSD • AVSD • Vibratory innocent murmur (Still's) • HOCM • Tricuspid regurgitation	• MR • Mitral stenotic murmurs • Mitral valve prolapse

Table 8.4: Sites of maximal intensity. AVSD, atrioventricular septal defect; HOCM, hypertrophic obstructive cardiomyopathy; MR, mitral regurgitation; PDA, patent ductus arteriosus; RVOT, right ventricular outflow tract; VSD, ventricular septal defect

3. Character: high- or low-pitched/musical/vibratory/blowing
4. Murmur-enhancing moves:
   - Left lateral position – brings out S3 and mitral murmurs
   - Sitting forward – brings out aortic murmurs
   - Change with inspiration/expiration – expiration brings out left-sided murmurs
   - Valsalva – for hypertrophic obstructive cardiomyopathy
5. Radiation (neck, axilla and back):
   - To neck – aortic in origin
   - To back/sides of chest – pulmonary in origin (eg pulmonary stenosis, branch pulmonary artery stenosis, right ventricular outflow tract obstruction/infundibular stenosis as in Fallot's)
   - To axilla – pulmonary valve murmurs or mitral regurgitation
   - Look for murmur over the back at this stage (for coarctation – just below tip of left scapula)
   - Remember innocent murmurs do not radiate
6. Grade (I–VI)

Grading of murmurs

I	Barely audible
II	Medium intensity
III	Loud but no thrill
IV	Loud with a thrill
V	Very loud but still requires stethoscope to be on the chest
VI	So loud can be heard with stethoscope off the chest

Auscultate basal lung fields before moving on.

Abdomen

- Hepatomegaly
- Situs inversus: associated with atrial isomerism; association with malrotation so may have evidence of Ladd's procedure scar
- Abdominal pacemaker: epigastric scar; may not be able to feel pacemaker box
- Scars from post-operative peritoneal dialysis (sometimes necessary for fluid/ascites management post bypass surgery)
- Ascites
- Evidence of protein-losing enteropathy (can occur after bidirectional Glenn procedure)
- Splenomegaly (occurs in infective endocarditis)

Groin

Inform the child that you are about to do this and remember to preserve modesty in older children:

- Scars from cardiac catheterisation/central lines
- Femoral pulses: compare character with upper limb pulses and look for radio-femoral delay (a sign that collateral circulation has developed to bypass an aortic coarctation)

Don't forget to mention

- Blood pressure if not already done
- Jugular venous pressure if not already done
- Femoral pulses if not already done
- Pulse oximetry
- Plotting growth chart
- Peripheral or sacral oedema
- Fundoscopy/dipping urine for evidence of infective endocarditis

8.3 Cases

The following are representative of some of the cases that you may see in the exam

Case one: Complex cyanotic congenital heart disease

Patient: 8-month-old girl. She appears small for her age although I would like to plot her on a centile chart. She is tachypnoeic at rest. She is centrally cyanosed. She has no dysmorphic features. She is clubbed and has peripheral cyanosis. She has a right-sided thoracotomy scar. On auscultation she has a normal first heart sound and a single second heart sound. She has a continuous murmur over the right side of her chest. (At the end of this examination you should suggest palpating the abdomen for liver and taking the blood pressure.)

This set of clinical signs would fit with a child who has a complex cyanotic congenital heart lesion and has had a shunt procedure.

Notes

Possible diagnoses include:

- Pulmonary atresia without a ventricular septal defect
- Double outlet right ventricle
- Univentricular heart with pulmonary atresia

You would not be expected to come up with that diagnosis, but carefully eliciting the clinical signs will allow a best guess. In this case, with the shunt and the single second heart sound, a diagnosis of pulmonary atresia would be the first best guess. Fallot's is less likely because there is no systolic murmur of pulmonary stenosis and in pulmonary stenosis, there should be two components of the second sound.

Case two: Trisomy 21, with AVSD and Eisenmenger syndrome

Sarah is a 7-year-old girl with features suggestive of Down syndrome. She is not cyanosed and is not clubbed. She has normal pulses with a rate of 90 bpm. Her apex beat is not displaced. She has a central sternotomy scar and what appear to be scars from chest drains. She has a systolic murmur heard loudest at the apex.

She has signs of mitral regurgitation following surgical repair of her AVSD.

I would like to plot her growth on a Down syndrome growth chart, and check her blood pressure to complete my examination.

Notes

You may be asked about the complications of AVSD if not corrected surgically (potential for pulmonary hypertension and Eisenmenger syndrome).

It is not necessary to list all the negative findings on examination, however if there are any that you feel are particularly pertinent to the case then do mention them.

Case three: Cavo-pulmonary shunt

Emma is a 20-month-old girl. She appears to be thriving although I would like to plot her on a centile chart. She has no dysmorphic features and appears to be comfortable at rest. She is mildly cyanosed centrally. Her pulses are normal throughout. She has no radio-femoral delay. She has a median sternotomy scar but no thoracotomy scars. Her apex beat is localised to the 5th intercostal space on the right; there are no heaves or thrills. She has normal heart sounds on the right side and appears to have dextrocardia.

This set of clinical signs would fit with a child who has a complex congenital cardiac defect and who is likely to have had a central shunt procedure, eg cavo-pulmonary anastomosis.

Notes

If the heart sounds are difficult to hear, as a matter of routine listen to the right side of the chest to exclude dextrocardia. Mild cyanosis in a child with a median sternotomy scar suggests a palliative shunt procedure for those children for whom definitive surgery is not possible. The child described had a complex cardiac defect involving right atrial isomerism, dextrocardia and a single ventricle with pulmonary atresia. A precise diagnosis would not be expected.

Case four: Innocent murmur

Lily is a 3-year-old girl who looks well. Shis is pink and not clubbed. She appears well grown for her age. She has normal pulses with a rate of 110 bpm. She has a 2/6 ejection systolic murmur, heard loudest in the pulmonary area, which does not radiate. The murmur is quieter when she sits forward. Her heart sounds are normal. Chest X-ray and ECG are normal.

I think Lily has an innocent flow murmur.

I would like to plot her growth on a growth chart and check her blood pressure to complete my examination.

Case five: Complex congenital cardiac disease

Jack is a 3-year-old boy. He looks well and is not tachypnoeic at rest. His right radial pulse is absent. He has a scar from cardiac catheterisation. He has a right thoracotomy scar and midline sternotomy scar. I presume he has complex congenital heart disease, now repaired, and I would like to proceed to listen to the heart.

It is not possible to achieve a specific diagnosis in this case.

Case six: Ventricular septal defect

Sam is a 7-year-old boy who looks well grown for his age. On examination he is not cyanosed or clubbed and has no respiratory distress. His pulses are normal with a rate of 75 bpm. There is a 3/6 pansystolic murmur that I can hear all over his praecordium. There is no radiation and no variation with posture or expiration. There are no other detectable heart murmurs. His heart sounds are normal; his apex beat is normal.

He has the clinical findings suggestive of a small ventricular septal defect. I would like to plot his growth on a growth chart and check his blood pressure to complete my examination.

Notes

If there is time the examiner may ask about treatment options. This depends on the size of the lesion and if the child is symptomatic. If the defect is small and the child is asymptomatic then a watch-and-wait with regular reviews approach is usually taken. Most small VSDs will close spontaneously.

Medical treatment is with diuretics +/– angiotensin converting enzyme (ACE) inhibitors.

Close attention should be paid to the child's growth and nutrition – they will often need high-calorie feeds.

Case seven: Supravalvular aortic stenosis

Luke is a 3-year-old boy who appears to be short for his age. He has some facial characteristics of Williams syndrome. He appears well and is pink. There is no clubbing or evidence of infective endocarditis in his hands. His radial and brachial pulses are normal and symmetrical in character. He has no chest wall deformities or scars. His apex beat is not displaced and is normal in character. He has a suprasternal and carotid thrill as well as a thrill over his aortic area. He has a loud, grade 4/6 ejection systolic murmur in the aortic area, which radiates to the carotids and is increased in expiration. He also has an ejection systolic murmur, grade 2/6, heard over the pulmonary area and widely radiating to the back and sides of the chest. This is not associated with a thrill. There is no ejection click or diastolic murmur. His heart sounds are normal. His femoral pulses are normal with no radio-femoral delay. There were no other abnormalities on cardiovascular examination.

My impression is that this boy has aortic stenosis, which, given the absence of an ejection systolic click and the features suggestive of Williams, I would expect to be a supravalvular aortic stenosis. He also has some evidence of branch pulmonary stenosis. I would now like to examine his blood pressure, look in his eyes for evidence of a stellate iris and examine his development.

8.4 Background notes

Measuring jugular venous pressure (JVP) in children

Measuring the JVP is a skill many of us may have forgotten or lost since working in adult medicine as it is rarely part of our routine examination. It can be time consuming, and it relies on co-operation and the presence of a neck; therefore, it is only normally attempted in older children (2 years and above). It can give useful information about fluid status in general and also about the condition of the right heart. Because it is time consuming it would be worth saying as part of your structured examination that you would normally measure the JVP and that you will come back to it at the end. That way if the examiner would like to see how you do it they can prompt you at this point. If they don't then you will need to remember to come back to it, as it may give you very useful information that you can use to inform your diagnosis.

Spend time getting the patient positioned exactly how you would like; doing this means that you are much more likely to be successful. Use pillows behind their head if necessary to get the sternocleidomastoid muscle completely relaxed. If you are not used to measuring JVP in children find a consultant (preferably a cardiologist or a nephrologist) to show you how they do it:

1. Position patient:
   - Head resting on bed – sitting up at an angle of approx 45°
   - Turn them slightly to the opposite side from where you are examining, this relaxes the sternocleidomastoid muscle
   - Ideally have the patient lit from the side
2. Locate pulsation: you can confirm that it is an internal jugular pulsation by:
   - Waveform (multiphasic waveform is normal for JVP; not seen in carotid pulsation)
   - Rises with pressure over the liver (ask if they have pain first); hepatojugular reflex
   - Non-palpable
   - Stops with pressure applied to the base of the neck
   - Position changes with raising/lowering the head
   - Varies with respiration
3. Measure height above sternal angle:
   - 4 cm above the sternal angle is quoted as within the normal range for adults

Characteristics of the JVP

The JVP pulsation consists of three waveforms:

- a-wave – corresponds to right atrial contraction
- c-wave – corresponds to ventricular contraction – causing pressure on the tricuspid valve as the atria are filling
- v-wave – corresponds to blood filling the right atrium before the tricuspid valve re-opening again

Associations between conditions and abnormal JVP findings

- Increased a-wave is seen in pulmonary hypertension
- Tricuspid regurgitation increases the v-wave
- Cannon a-waves are seen when the atria are contracting against a closed tricuspid valve (complete heart block)
- Raised JVP seen in constrictive pericarditis/cardiac tamponade (unlikely exam case), right-sided heart failure

Innocent murmurs

Features

1. Asymptomatic
2. Normal heart sounds
3. Systolic or continuous (never solely diastolic)
4. Do not radiate
5. Vary with position and respiration
6. Soft (grades 1–3)
7. Normal ECG and chest X-ray

Some examples of innocent murmurs

1. Pulmonary flow murmur:
    - Very common
    - Characteristically it is brief and in mid-systole
    - Loudest with the patient lying down and during expiration
    - Occurs in children and adolescents of all ages
    - Commonly heard during hyperdynamic states such as fever, anaemia, thyrotoxicosis
2. Still's murmur:
    - Typically aged 3–8 years, resolves towards adolescence
    - Grades 1–3 present in early systole, heart sounds normal
    - Maximum intensity between lower left sternal edge and apex
    - Vibratory in nature

   - Reduces in intensity/disappears when patient stands up/carries out Valsalva manoeuvre
3. Venous hum:
   - This is the only innocent murmur that has a diastolic component
   - High-frequency continuous murmur most commonly heard in children aged 2–6 years
   - Best heard over the supraclavicular fossa on the right with the head turned to the other side
   - May radiate and is often heard on both sides
   - Sometimes an associated thrill
   - Disappears on lying flat or if the neck veins are compressed

Dextrocardia

- There is no excuse to miss dextrocardia in the exam. Cases are quite common and readily available to the examining centre to put into the exam to catch people out. If you have any difficulty locating the apex either on palpation or auscultation it is worth auscultating the right side of the chest. It is probably sensible to include it in your examination as a matter of routine to show to the examiner that you remember to look.
- Dextrocardia can be isolated or in association with situs inversus. Dextrocardia without situs inversus can be associated with almost any cardiac malformation! (Single ventricle, transposition of the great arteries, pulmonary stenosis/atresia, atrial septal defects, ventricular septal defects, atrioventricular septal defects, total anomalous pulmonary venous drainage tricuspid atresia.)
- Dextrocardia in association with situs inversus is effectively when the heart and abdominal organs are mirror images of what they would be normally. Often despite this, the heart functions completely normally.
- Abnormalities of the position of the heart are also associated with asplenia and polysplenia syndromes.
- Dextrocardia with situs inversus can be a part of Kartagener syndrome – incidence 1:40 000, associated with primary ciliary dyskinesia and all the associated complications of this.
- If you find dextrocardia on examination, it is important that you go on to palpate the abdomen to find the position of the liver and to listen to the lung fields for bronchiectasis.

Ventricular septal defect

Pathophysiology

Ventricular septal defects (VSDs) are the commonest congenital heart defects, with a frequency of 1:500. Defects usually occur in the membranous part of the septum with extension into the muscular septum, the so-called perimembranous VSD, although they may occur anywhere. The main haemodynamic effect is left ventricular volume overload and increased pulmonary blood flow. There is often a misconception that the right ventricle is overloaded due to the left-to-right shunt of blood. When the blood is being shunted in systole, the right ventricle is ejecting blood into the pulmonary artery, so the blood is shunted directly into the pulmonary artery and hence into the lungs. This then returns to load the left ventricle during diastole.

Investigations

ECG – will be normal with a small defect; in large defects signs of biventricular hypertrophy develop from a few months of age. Chest X-ray – again may be normal with a small defect, but may show cardiomegaly +/– increased pulmonary markings in larger defects. Echocardiogram – can help estimate the size of the shunt by examining the degree of volume overload of the left atrium and left ventricle.

Presentation

In a small defect where the shunt is insignificant, there is usually no haemodynamic derangement, and the patient is asymptomatic. An ECG will be normal. A chest X-ray will also be normal. A large defect producing a shunt with haemodynamic significance may cause signs of heart failure, with cardiomegaly, a diastolic murmur at the apex due to increased blood flow into the left ventricle from pulmonary veins in diastole and a loud second heart sound due to an increase in pulmonary blood flow. The ECG may show a left ventricular volume overload or biventricular hypertrophy with left ventricular hypertrophy predominant if the defect is large enough. Chest X-ray may show cardiomegaly, enlarged pulmonary arteries and increased vascular markings from pulmonary oedema, again if the defect is large.

Diagnosis

The diagnosis of a VSD is made on echocardiography. As 80% of the defects close spontaneously, a reassuring line can be taken.

Treatment

- In haemodynamically significant defects, treatment is indicated. Medical management aims to control congestive cardiac failure and to prevent the development of pulmonary vascular disease, with the maintenance of normal growth using calorie supplementation if appropriate. This involves the use of diuretics and angiotensin converting enzyme (ACE) inhibitors such as captopril.

- Indications for surgical closure of VSD – surgical management is reserved for those with a significant shunt who are not responding to medical management. The size of the intracardiac shunt is usually expressed as the ratio of pulmonary to systemic blood flow. If the cardiac shunt is small, ie a pulmonary to systemic flow ratio (Qp:Qs) of less than 2:1, the cardiac chambers are unlikely to be enlarged and the pulmonary bed is likely to be normal. If the shunt is large with a pulmonary to systemic flow ratio greater than 2:1, left atrial and ventricular volume overload occurs and there is significantly raised pulmonary arterial pressure leading to pulmonary hypertension. This will ultimately lead to irreversible damage and the development of Eisenmenger syndrome (see later). Cardiac surgery is usually considered if the pulmonary to systemic flow ratio is persistently greater than 2:1. In complicated cases of patients who are not suitable for early surgery, pulmonary artery banding is sometimes carried out, eg multiple VSDs – 'Swiss cheese' defect. Infants who require medical management will normally be repaired before 6 months (before 3 months in trisomy 21). For a moderate defect, if growth is normal and no medical management is required, they may have surgical repair aged 2–5 years. For very small defects, watch-and-wait (may close spontaneously).

Examination finding in asymptomatic VSD

This is one of the commonest short cases.

Inspection

- Normal, patient appears well
- No tachypnoea or tachycardia
- Hands – no clubbing/cyanosis
- Pulses normal
- Blood pressure normal
- Normal face
- No scars

Palpation

- Position and character of apex beat are important in determining the haemodynamic significance of VSD
- Active praecordium – an indication of volume loading secondary to left-to-right shunt – unlikely to find this in an older child (as most haemodynamically significant VSDs present with symptoms and are corrected in infancy)
- Thrill, normally at the left sternal edge

Auscultation

- ○ Pansystolic murmur, maximal at the left sternal edge
- ○ No radiation, but heard all over the praecordium
- ○ Specifically mention second heart sound
- ○ A loud second heart sound suggests elevated pulmonary arterial pressure, ie a haemodynamically significant VSD causing pulmonary hypertension, therefore you must mention the second heart sound
- ○ A diastolic rumble at the apex or any clinical features of heart failure imply a large defect with a significant shunt
- ○ When presented with what you feel is a simple VSD in the exam you must include a statement about whether or not it is likely to be haemodynamically significant in your summary

Features to look for in a haemodynamically significant VSD

- Tachypnoea/symptoms
- Murmur may be softer (implying larger lesion)
- Hyperactive praecordium
- Loud second heart sound (has already developed complications)
- Displaced apex beat
- Diastolic murmur
- Effects on growth

Supplementary questions frequently asked

- ○ How would you confirm this is a VSD?
- ○ How would you follow this child up?
- ○ What advice would you give the parents?
- ○ Give the options for management
- ○ What are the indications for surgery?
- ○ How would you differentiate between VSD and an innocent murmur?

Atrial septal defect

Pathophysiology

The most common site (80%) of the defect is in the fossa ovalis (ostium secundum). Other sites include the septum primum (ostium primum defect, more recently called partial atrioventricular septal defect or partial AVSD) and the atrioventricular septum (atrioventricular septal defect, atrioventricular canal defect, more recently called complete AVSD). The main haemodynamic abnormality is right ventricular volume overload during diastole with an increased pulmonary blood flow secondary to the shunt. Most children are asymptomatic and the lesion is discovered on routine examination. Symptoms of an ASD rarely develop before the third decade. Patients may then go on to develop atrial arrhythmias, pulmonary hypertension and heart failure.

Investigations

- ECG:
  - Ostium secundum: right axis deviation, partial right bundle branch block
  - Ostium primum (also called partial endocardial cushion defect, part of a spectrum that can include AVSD with involvement of atrioventricular valves). Occurs in the lower part of the septum: left axis deviation, partial right bundle branch block
- Echocardiogram

Treatment

The majority of small (ie < 6 mm) isolated ASDs will close spontaneously in the first few years of life. Treatment is usually surgical closure around the third year of life, or earlier if symptomatic. If left untreated, the defect will cause symptoms in adult years. Currently a child of 3 or over may be suitable for repair using a transcatheter closure device. The procedure is considered to be low risk. Antibiotic prophylaxis is required for surgical and dental procedures for 6 months following repair. Bypass surgery may be required for larger defects or defects which are unusually shaped (and therefore not suitable for device closure).

Examination findings in ASD

General

- Normally children with ASD are asymptomatic: they should be well grown, not clubbed, etc

Palpation

- Apex beat normal, may have parasternal heave, no thrills

Auscultation

- Heart sounds both heard
- Second heart sound is widely split, with fixed splitting due to overload of the right ventricle with prolonged ejection into the pulmonary arterial circulation
- Ejection systolic murmur upper left sternal edge (alternatively there may be no murmur). The ejection systolic murmur is due to increased flow across the right ventricular outflow tract. It is normally grade 2–3/6
- May be a mid-diastolic murmur due to high flow across the tricuspid valve (lower left sternal border), which is an excellent diagnostic sign. Occurs when there is a large left-to-right shunt

Supplementary questions frequently asked

- ❍ How would you confirm the diagnosis?
- ❍ How would you manage the patient?
- ❍ What are the indications for surgery?

Atrioventricular septal defect

Pathophysiology

Atrioventricular septal defects (AVSDs), also known as endocardial cushion defects, represent a spectrum of cardiac anomalies. A very mild AVSD may consist of just an ostium primum ASD. Worsening severity is associated with an increasing degree of clefts in the atrioventricular valves and ventricular septal defects, in addition to the ASD.

AVSDs represent 4% of congenital cardiac anomalies. There is a strong association with Down syndrome; 40% will have congenital heart disease, of these approximately 40% will have an AVSD.

There is left-to-right shunting through the atrial or ventricular defect. If this is not treated it leads to atrial dilation, arrhythmias, volume-loaded ventricles and pulmonary hypertension.

Investigations

- ❍ On ECG, AVSD classically causes a 'superior axis', which means that the axis is between –40 and –150 degrees. One would also expect to see right ventricular hypertrophy, a prolonged PR interval and some right-sided conduction delay
- ❍ Chest X-ray may show cardiomegaly and pulmonary plethora
- ❍ Echocardiogram

Treatment

Medical treatment is usually short term to manage/prevent cardiac failure with diuretics +/– ACE inhibitors and to optimise the child's nutritional status and clinical condition before surgery.

The mainstay of treatment is surgical. Complete repair, ie closing all septal defects and repair of the AV valve is normally carried out within the first 6 months of life. This is to prevent the development of pulmonary hypertension. Children with Down syndrome are at particular risk of pulmonary hypertension, and are likely to undergo surgical correction at a younger age.

Potential post-operative complications

- Damage to AV node/conduction fibres
- Ongoing AV valve dysfunction
- Problems with increased pulmonary vascular resistance

Examination

General points

Look for AVSD in any child with Down syndrome, but remember that other cardiac lesions are common in this group as well (see below). If there are clinical signs consistent with mitral regurgitation, one can confidently conclude that the most likely lesion is an AVSD.

Signs of heart failure are common before correction, and it is important that you comment on the presence or absence of heart failure if you suspect an AVSD. The development of pulmonary hypertension is a key factor in determining the timing of surgery. This is associated with a loud S2, and it is crucial that you comment on whether or not there is a normal second heart sound.

You are more likely to see these patients in the exam after they have had a repair (ie with a median sternotomy scar). Look specifically for signs of residual mitral regurgitation, and if these are present, comment on whether you think this is significant by looking for cardiac enlargement, cardiac failure or pulmonary hypertension.

Before surgical correction

General

- May have features of Down syndrome
- Comment on growth (remember to refer to the Down syndrome growth charts)
- Look for signs of increased work of breathing at rest (tachypnoea, head bobbing, wheeze)

Chest

- Inspection (look for hyperactive precordium and intercostal recession)
- Palpation – hyperdynamic apex beat if in cardiac failure, right ventricular heave from right ventricular volume loading, palpable thrill at lower left sternal edge (LLSE)
- Auscultation – loud pansystolic murmur at LLSE, also mitral regurgitation murmur at apex. Comment on the character of the second heart sound. May hear gallop rhythm if in failure. May hear mid-diastolic murmur from flow across AV valves

Other

- Look for hepatomegaly
- Be prepared to comment on the likely ECG findings, treatment of cardiac failure (diuretics, ACE inhibitors, nutrition) and subsequent investigations (echo/cardiac catheter, chromosomes)

After surgical correction

- Comment on growth
- Look for signs of residual mitral regurgitation (regurgitant murmur at the apex radiating to the axilla, diastolic flow murmur at the apex, gallop rhythm may be present)
- Comment on surgical scars

Signs of heart failure

- Faltering growth
- Tachycardia
- Increased work of breathing
- Cardiomegaly (displaced apex beat)
- Hyperdynamic apex beat
- Gallop rhythm
- Basal crepitations, wheeze or other signs of alveolar fluid
- Hepatomegaly
- Sacral/peripheral oedema

Likelihood of cardiac lesions in Down syndrome

- 40%–50% will have cardiac anomalies
- 40% will have AVSD
- 30% will have VSD
- 10% will have ASD (secundum)
- 6% will have tetralogy of Fallot
- 4% will have PDA
- 30% will have more than one anomaly

Aortic regurgitation

Pathophysiology

Associated syndromes – Ehlers–Danlos syndrome, Marfan syndrome, Turner syndrome. Acquired causes – rheumatic fever, systemic lupus erythematosus. Significant aortic regurgitation leads to left ventricular volume overload and left ventricular hypertrophy. This can lead to increased left atrial pressures and pulmonary oedema. Children with aortic regurgitation may be asymptomatic for many years. They may then present with decreased exercise tolerance and dyspnoea.

Investigations

- ECG may show left ventricular hypertrophy
- Chest X-ray may show cardiomegaly
- Echocardiography

Treatment

This involves surgical correction with valve replacement before irreversible left ventricular damage occurs. Surgical correction is indicated for exercise-induced symptoms. The patient would normally need to have anticoagulation after valve replacement

Clinical findings

- Look for associated conditions
- Look for water-hammer pulse in radial artery (bounding radial pulse when arm is elevated)
- Collapsing carotid pulse (Corrigan's pulse), wide pulse pressure
- High-pitched decrescendo murmur along the left sternal edge in diastole. This is best heard with the patient sitting up, leaning forward, in expiration. It may be associated with a thrill
- Soft S1
- Signs of left ventricular dilation (displaced hyperdynamic apex beat)
- S3 may be heard because of reduced left ventricular compliance
- Austin Flint murmur (mid-diastolic rumble caused by the effect of regurgitant volume of blood on flow across mitral valve) may be heard

Pulmonary stenosis

Pathophysiology

- Present in 5%–8% of children with congenital cardiac disease: valvular, subvalvular, supravalvular
- Supravalvular – rare, associated with congenital rubella or Williams syndrome

- Peripheral pulmonary artery stenosis – associated with congenital rubella or Alagille syndrome
- Haemodynamically, pulmonary stenosis causes pressure overload of the right ventricle and a relatively fixed cardiac output

Presentation

- Patients are usually asymptomatic if the stenosis is mild or moderate
- Neonates can get critical pulmonary stenosis with cyanosis due to shunting across the foramen ovale. This is a separate entity and requires intravenous prostaglandin to keep the ductus arteriosus patent and urgent surgery

Investigations

- ECG: usually normal; in severe stenosis there may be evidence of right ventricular hypertrophy and right axis deviation
- Chest X-ray: usually normal, although you may see a prominent pulmonary artery due to post-stenotic dilatation. May see prominent right atrium and right ventricle
- Echocardiography: diagnostic

Treatment

- Surgery is only indicated in supravalvular or subvalvular pulmonary stenosis or if valvuloplasty is unsuccessful
- Balloon valvuloplasty may be done
- Remember, in the exam children are often seen post repair
- Post repair the child is likely to have a residual systolic and diastolic murmur (secondary to pulmonary incompetence). There may be a midline sternotomy following complete repair, or no scar (balloon valvuloplasty).

Examination findings in pulmonary stenosis

Inspection

- Most likely to be asymptomatic

Palpation

- May have thrill over pulmonary area
- May have right ventricular heave (uncommon)

Auscultation

- Ejection systolic murmur maximal at the upper left sternal edge, radiation through to the back
- If it is a critical stenosis there may be no murmur; it will present in the neonatal age group but outside of this age group, louder and longer murmurs mean more severe stenosis
- Heart sounds both heard: widely split second heart sound, with a softer pulmonary component, can even be absent if the stenosis is severe
- Ejection click

Further examination

- Usually normal but look for the stigmata of Noonan syndrome or Williams syndrome
- Look for evidence of right ventricular failure. This is unlikely as the lesion is likely to have been repaired before this stage, but you will need to show the examiners that you are aware that this is a complication of pulmonary stenosis.

Aortic stenosis

Pathophysiology

- Five per cent of congenital heart disease: valvular, subvalvular, supravalvular:
  - Valvular (most common): usually valves are abnormal with one or two cusps (bicuspid) rather than the usual tricuspid. Valvular aortic stenosis is often associated with coarctation of the aorta
  - Supravalvular: this is a diffuse/localised narrowing immediately above the aortic sinuses and the coronary arteries. Most commonly associated with Williams syndrome. Many patients also have peripheral pulmonary artery stenosis in addition
  - Subvalvular: long narrowing of the left ventricular outflow tract
  - Subaortic stenosis: this is a fibrous diaphragm just below the aortic valve, often associated with an abnormal subaortic region. It is often seen with coarctation. Clinical presentation is similar to that of aortic stenosis although it is often not severe. There is no ejection click. Treatment consists of excision of the fibrous diaphragm in all but the mildest of cases as it can be easily performed without interfering with other structures. Subaortic stenosis patients are particularly prone to bacterial endocarditis
- The left ventricle becomes hypertrophied due to outflow obstruction. There is effectively a fixed cardiac output
- Symptoms can include dizziness, palpatations, chest pain and even syncope on exertion, although most patients are asymptomatic
- Syncope is important as it indicates severe disease

Investigations

- ECG is normal in mild obstruction. In severe lesions there may be left ventricular hypertrophy on the ECG, but it is often surprisingly normal:
  - ECG signs of left ventricular hypertrophy:
    - Tall R waves V5–V6
    - Inverted T waves in 1, aVL, V5–V6
  - ECG signs of ischaemia (in very severe lesions):
    - ST depression
    - T wave inversion

- Chest X-ray – may be normal or show a prominent left ventricle
- Echocardiography is always indicated. It is diagnostic and will assess the gradient across the stenosis

Treatment

Treatment is conservative in most cases, avoiding valve replacement in the young patient. If the gradient across the valve is greater than 60 mmHg, intervention is indicated. This is usually in the form of a balloon valvuloplasty by cardiac catheter, or surgical valvuloplasty. Aortic regurgitation can develop after balloon procedures, which often needs surgical correction.

As with pulmonary stenosis, the patient may present post repair, with the murmur of aortic stenosis still present and the murmur of aortic regurgitation due to the residual incompetence of the valve post-operatively. If the patient is older, they may have had the stenosis repaired via an open valvuloplasty and could have a median sternotomy scar. Younger children may have had a catheter repair and will therefore not have a sternotomy scar.

Associations with aortic stenosis

- Turner syndrome
- Williams syndrome (supravalvular aortic stenosis)
- Coarctation of the aorta (particularly valvular aortic stenosis with bicuspid aortic valve)
- Other cardiac abnormalities, eg hypoplastic left ventricle, mitral valve abnormalities, HOCM

Examination findings in aortic stenosis

Take care to note associated aortic regurgitation findings if the child has had a procedure to relieve gradient across aortic stenosis.

Inspection

- Normally asymptomatic
- Look for dysmorphic features (Turner/Williams in particular)

Pulse

- May be collapsing (if associated with aortic valve incompetence post procedure to open stenotic area), although usually normal
- If the defect is severe, pulse pressure may be low, causing weak peripheral pulses

Blood pressure

- Normal or slightly raised, narrow pulse pressure
- In supravalvular aortic stenosis, it is possible to get a higher blood pressure reading in the right compared to the left arm

Palpation

- Suprasternal and carotid thrill
- May also have thrill over aortic area (upper right sternal edge)
- Apex beat may be displaced and forceful

Auscultation

- Heart sounds may be normal
- In severe stenosis, quiet A2
- Ejection click heard loudest at lower left sternal edge and apex
- Ejection systolic murmur at the lower left sternal edge, in the aortic area and radiating into the neck – 'harsh' sounding
- Early diastolic murmur (decrescendo) if valvular aortic stenosis with aortic regurgitation

Further examination

- Ask to feel femoral pulses and feel for radio-femoral delay because of association with aortic stenosis
- Listen at the back to confirm (aortic stenosis does not radiate to the back – coarctation does)

Coarctation of the aorta

Pathophysiology

This is present in 8%–10% of patients with congenital heart disease.

In 98% of cases it originates just below the origin of the left subclavian artery, at the origin of the ductus arteriosus – the juxtaductal coarctation. The remaining ones are pre-ductal and often associated with arch hypoplasia. The male: female ratio is 2:1. Pre-ductal is more serious and often presents in the neonatal period due to its severity. The juxtaductal type may be mild and often does not present until late childhood. Even then it is often asymptomatic and comes to light during the investigation of hypertension.

The classical signs are of a disparity in pulses and blood pressure between the arms and the legs, hence the pulses in the arms and legs should always be palpated together at some point in a cardiovascular examination. The delay occurs due to the fact that the blood flow to the descending aorta is dependent on collaterals, hence the femoral pulse is felt after the radial. The blood pressure difference should be measured in all four limbs. Normally, the systolic pressure in the legs is 10–20 mmHg higher than in the arms, in coarctation it is lower. If the BP is higher in the right arm than in the left, it suggests involvement of the left subclavian artery in the coarctation.

Investigation

- Chest X-ray: usually normal in the juxtaductal coarctation unless it does not present until after the first decade, when there may be mild to moderate cardiac enlargement due to left ventricular prominence. Rib notching occurs in children with long-standing coarctation (> 7 years).
- ECG: usually normal unless late presentation as above, when there may be signs of left ventricular hypertrophy.
- Echocardiography: demonstrates the presence and position of the coarctation. Will also determine the pressure gradient across the coarctation and exclude an intracardiac abnormality.

Surgical treatment

A subclavian flap, Gore-tex® graft or end-to-end anastomosis can be used to repair the abnormal segment. Repair is through a left-sided thoracotomy (if aortic arch is on the normal side!). Complications include residual hypertension and re-coarctation. The latter can be treated with balloon dilatation, although occasionally further surgery is required. Subclavian flap repair causes the loss of the pulse in the left arm. Older children (> 10) can be treated by insertion of a stent during cardiac catheterisation.

Examination findings in coarctation of the aorta

It is very unlikely you will have a patient in the exam who has untreated aortic stenosis.

Inspection

- Look for dysmorphic features (Turner syndrome)
- Check blood pressure

Pulses

- Right radial pulse normal
- Compare radial pulses; left radial may be absent
- Femoral pulses may be weak
- May be radio-femoral delay

Scars

- None unless there has been a repair. It is likely that if you see a case in the exam it will be post repair (left thoracotomy scar)
- If the scar is associated with a reduced left radial pulse volume, think subclavian flap repair

Palpation

- Apex beat is usually normally placed, no heaves or thrills

Auscultation

- Systolic murmur is loudest at the back (left interscapular area). Alternatively the murmur may be non-specific or absent
- Ejection click at the upper left sternal edge if there is a bicuspid aortic valve
- There may be a diastolic murmur from collaterals in the older patient

Further examination

- Could be directed at looking for evidence of Turner syndrome
- Offer to do four-limb blood pressure measurement

Associations

- Turner syndrome
- Bicuspid aortic valve in 70%
- Over half of the patients with coarctation have other lesions, eg ventricular septal defect, mitral valve abnormalities and aortic stenosis. Associated abnormalities are more likely if coarctation is symptomatic in infancy

The cyanosed child

Cases in the exam are often quite difficult. Many will have had surgery and the cardiac lesions are likely to be complex. It is important to describe what you see and to make sensible statements about potential causes. It is therefore essential to have a good overview of cyanotic congenital heart disease. Some useful information is summarised below. While thinking about this it is important to remember that children seen in the exam who have had definitive surgery for cyanotic congenital heart disease will be pink (eg transposition of the great arteries or tetralogy of Fallot) and will present as a pink child with a central sternotomy scar with, in some cases, an additional shunt scar. Children seen in the exam who have cyanosis due to cardiac causes are likely to have complex cyanotic congenital heart disease.

Cardiac causes of cyanosis

- Decreased pulmonary blood flow, eg tetralogy of Fallot, pulmonary atresia, Ebstein's anomaly, tricuspid atresia
- Poor mixing, eg transposition of great arteries
- Common mixing, eg truncus arteriosus, double outlet right ventricle, total anomalous pulmonary venous drainage, univentricular heart. Cyanosis in these conditions may be mild enough to be difficult to detect clinically

Management

Fallot's tetralogy

See below for details on how to manage this condition.

Pulmonary atresia

Pulmonary atresia with or without an intact intraventricular septum is a difficult problem, and usually requires several shunt procedures; definitive repair is often not possible. Often there is a poor prognosis. Blood supply to the lungs may be provided by major aortopulmonary collateral arteries (MAPCAs), which may cause pulmonary oligaemia or pulmonary oedema depending on their size and distribution.

Treated by unifocalisation, which involves fashioning a main pulmonary artery-like structure out of whichever vessels best suit. This can then be incorporated into a Glenn or Fontan circulation if the right ventricle is small. If the right ventricle is normal or near normal in size and function then repair would involve attaching a graft which links the right ventricle to the newly fashioned pulmonary arteries, bypassing the atretic pulmonary valve (Contegra graft).

Transposition of the great arteries

Transposition of the great arteries is the second commonest cyanotic lesion, comprising 4% of all congenital disease. Infants at presentation need a prostaglandin infusion, followed by urgent transfer to a cardiology unit. The arterial duct is maintained by a prostaglandin infusion and a definitive repair is carried out within the first few days (arterial switch). If there is likely to be a delay in definitive surgery, or if there is inadequate mixing through the ductus, an atrial septostomy may be performed.

Truncus arteriosus

Truncus arteriosus is rare, comprising 0.5% of all lesions. Often associated with chromosomal abnormality (22q deletion syndrome).

Double outlet right ventricle

Double outlet right ventricle is rare. A very heterogeneous group of conditions. Early pulmonary banding (if pulmonary blood flow is high) or shunt procedure (if pulmonary blood flow is inadequate) followed by definitive repair at a later date.

Total anomalous pulmonary venous drainage

Total anomalous pulmonary venous drainage is rare, comprising 1% of all cardiac cases (40% have other associated cardiac lesions). Definitive repair early for obstructive lesions.

Univentricular heart

Univentricular heart is as common as transposition: 4% in certain published series. Many different types. There is no correction, only palliation. Children with this condition often die early. If cyanosis is severe, may need shunt procedure early. Pulmonary artery banding if the pulmonary artery flow is high. If the pulmonary outflow tract and pulmonary arterial tree is underdeveloped the patient may need a palliative shunt procedure, which will encourage the vessels to grow. New shunts may be needed as the patient grows, or a Glenn procedure (superior vena cava to pulmonary artery). If they reach 3–5 years, surgery to palliate the defect is indicated, involving the Fontan/modified Fontan procedure (total cavo-pulmonary anastomoses), bypassing the ventricle.

Hypoplastic left heart syndrome

Hypoplastic left heart syndrome makes up 8% of all congenital heart disease. It is often grouped with univentricular heart disorders. It occurs when the left-sided heart structures (ie left ventricle, mitral and aortic valves and the first section of the aorta) are significantly underdeveloped. As a result, blood flow from the left ventricle to the aorta is minimal, and therefore the presence of a patent ductus arteriosus is essential to maintain the peripheral circulation.

Treatment is a succession of staged palliative surgeries individual to the child's anatomy. Usually this is in the form of a Norwood, followed by a bidirectional Glenn procedure and finally a Fontan. Long-term prognosis is difficult to predict as up to 40% of children will not survive the surgeries. At present there are children in the UK who have survived into their teens. Heart transplantation can be considered; though this is usually only considered in older teens.

Fallot's tetralogy

Pathophysiology

Fallot's tetralogy forms 10% of congenital heart disease and is the most common cyanotic lesion beyond infancy (frequency 1:2000 births). It is associated with chromosomal abnormalities such as 22q11 deletion and Down syndrome.

The four constituents of Fallot's tetralogy are:

1. Right ventricular outflow obstruction (could be infundibular stenosis, pulmonary valve stenosis or both).
2. Ventricular septal defect.
3. Overriding aorta.
4. Right ventricular hypertrophy.

Investigations

○ Hb/packed cell volume if cyanosed

○ Chest X-ray: 'boot-shaped heart' with a prominent right ventricle, small pulmonary arteries, oligaemic lung fields

○ ECG: right axis deviation, right ventricular and right atrial hypertrophy

○ Echocardiogram: demonstrates the lesion and the size of the pulmonary arteries as well as the side of the aorta, obviating the need for cardiac catheterisation

Treatment

Corrective surgery, ie repair of VSD, and augmentation of right ventricular outflow tract is usually carried out within the first year of life. The timing depends on the severity of the right ventricular outflow tract obstruction, the clinical severity of the lesion, including degree of hypoxia, and the size of the child. Palliative shunt procedures (eg Blalock–Taussig shunt) are still performed in inoperable cases or to bridge patients until full repair can be done.

Complications

- Exertional dyspnoea – squat for relief
- Cerebral thrombosis/brain abscess
- Infective endocarditis
- Arrhythmias (in years following repair)
- Paroxysmal hypercyanotic spells:
  - The infant becomes restless, irritable and cyanotic
  - Rapid deep breathing
  - Decreased intensity of murmur with increasing cyanosis – decreasing conscious level
- The attacks are most common after exertion and during the first 2 years of life
- Treatment of paroxysmal hypercyanotic spells:
  - Bring baby's knees to their chest (reduces venous return and increases systemic resistance, reversing the shunt)
  - The following medications should only be given with tertiary cardiology input, and aim to improve pulmonary blood flow:
    - Morphine
    - Sodium bicarbonate if acidotic
    - Vasoconstrictors
    - Beta blockers

Examination findings in Tetralogy of Fallot

This is a classic short case and frequently seen. It will usually have been operated on by the time the child is seen.

Inspection

- Untreated, most patients are symptomatic – cyanosed/clubbed
- May have no cyanosis in milder forms
- Poor weight gain
- Hands: early clubbing
- Pulse:
  - Normal

- May be absent left-sided pulse in association with left thoracotomy scar (or a similar situation on the right)

❍ Face: central cyanosis (if uncorrected)

❍ Scars:
  - If untreated, none
  - May be left/right thoracotomy scar in association with an absent pulse on the ipsilateral side
  - There may be bilateral thoracotomy scars suggesting the failure of one shunt and the need for a second shunt procedure
  - Central sternotomy scar indicates that definitive repair has been carried out – the child will not be cyanosed (but may still have signs of right ventricular outflow tract stenosis)

Palpation

❍ Apex beat normally located

❍ Right ventricular impulse may be palpable

❍ Systolic thrill palpable at the upper left sternal edge in 50%

Auscultation

❍ Loud (grade 3–5/6) ejection systolic murmur maximal at upper left sternal edge, transmitted widely, especially to the lungs – radiates to the back

❍ Shunt murmur in younger children (you wouldn't expect to hear this if there is a median sternotomy scar, as indicates definitive repair) on the side of the thoracotomy

❍ Heart sounds: second heart sound is either single or the pulmonary component is soft

❍ May have an aortic ejection click

Eisenmenger syndrome

Pathophysiology

This is when a large left-to-right shunt over time causes increased pulmonary blood flow; this leads to increased pulmonary vascular resistance and a subsequent reversal of flow, ie a right-to-left shunt.

The most common lesions that cause Eisenmenger syndrome are ASD, VSD, AVSD and PDA. When Eisenmenger occurs the defect is usually large, although children with Down syndrome and ventricular septal defect or atrioventricular septal defect are particularly liable to develop pulmonary hypertension and reversal of shunt at an earlier age.

Presentation and investigations

❍ Central cyanosis with clubbing

❍ Pulse is of normal volume

❍ Right ventricular heave

- ○ Short ejection systolic murmur (ejection of blood into the dilated pulmonary artery) often preceded by an ejection click
- ○ Loud S2

Chest X-ray – right ventricular prominence with small peripheral pulmonary vessels (giving a 'peripheral pruning' appearance).

ECG – shows peaked 'P' waves due to right atrial hypertrophy, right axis deviation and moderate or marked right ventricular hypertrophy with tall R waves in V4R and VI.

Echocardiogram – will provide information on right ventricular function, pulmonary artery pressures and the underlying structural cause.

Treatment

Treatment is difficult; surgical closure is not possible. Heart–lung transplant is the only life-saving procedure. Medical treatment involves use of pulmonary vasodilators (eg sildenafil).

Cardiomyopathy

Four different categories – dilated and hypertrophic are the most common, others are restrictive and arrythmogenic right ventricular cardiomyopathy.

Hypertrophic cardiomyopathy

- ○ Abnormally thick and stiffened heart muscle, which cannot fully relax and allow normal ventricular filling. This leads to decreased cardiac output
- ○ Usually familial
- ○ Onset of symptoms very variable – decreased exercise tolerance/fatigue, cough, arrhythmias (risk of sudden death highest in this category of myopathies)
- ○ Clinically, may have symptoms of left ventricle failure in end-stage disease.
- ○ Chest X-ray – may be normal or show left ventricular or left atrial enlargement +/– right atrial enlargement
- ○ Murmur may be ejection systolic murmur from left ventricular septal hypertrophy or murmur from mitral regurgitation secondary to abnormal movement of the anterior leaflet
- ○ Treated medically with β-blockers, anti-arrhythmics
- ○ If surgery is required a myectomy can be performed. Occasionally patients may also need surgery to mitral valve

Dilated cardiomyopathy

- ○ Most common cardiomyopathy (60% of cardiomyopathies)
- ○ Causes: genetic/infections or environmental
- ○ History of a preceding viral illness in 50%

- Causes dilation and systolic dysfunction of all four chambers (usually left side worse than right)
- Onset of symptoms often insidious:
  - Cough
  - Poor feeding/sweating with feeding/poor weight gain
  - Shortness of breath
- Chest X-ray – may show massive cardiomegaly with pulmonary oedema
- Managed medically (diuretics/digoxin/ACE inhibitors/β-blockers) – may require cardiac transplantation
- Clinical findings may include:
  - Displaced apex beat
  - Mitral regurgitation or tricuspid regurgitation murmur
  - Third heart sound
  - Signs of left heart failure
  - Signs of right heart failure (including raised JVP)

Infective endocarditis

Normally requires predisposition of structural heart abnormality resulting in turbulence/pressure gradients, leading to platelet aggregation/potential for thrombus. Following bacteraemia, this abnormal area can become infected, resulting in vegetation formation. Bacteraemia is often associated with diseased teeth and/or dental procedures.

Signs of endocarditis

- Fever
- Osler's nodes: painful, red/raised lesions on fingertips (from immune complex deposition)
- Janeway lesions: non-tender, erythematous/haemorrhagic macules or nodules on palms or soles
- Splinter haemorrhages: dark red linear streaks in nail bed
- Conjunctival/buccal petechiae
- Clubbing
- Hepatosplenomegaly (in 20%)
- Congestive cardiac failure (depending on severity of valvular disease)
- Murmur (20%–80% of patients)
- Roth's spots: retinal haemorrhages with pale centre, seen on fundoscopy
- Microscopic haematuria
- Neurological abnormalities
- Look at dentition/gingival disease

Endocarditis prophylaxis

Previously all patients with any type of congenital heart defect would receive antibiotic prophylaxis. However antibiotics are now only recommended for patients at high risk of infective endocarditis:

- A prosthetic valve/valve repair with prosthetic material
- A history of endocarditis
- Congenital cyanotic heart disease
- Surgically repaired atrial septal defect/ventricular septal defect/patent ductus arteriosus – for 6 months after surgery
- Surgically repaired congenital heart disease with abnormal flow adjacent to any prosthetic material

Patients should be counselled by their cardiologist as to whether they require prophylaxis and carry information with them about what regime is appropriate.

Syndromes associated with cardiac lesions

Table 8.5 is a list that may help when faced with a dysmorphic child in the cardiac station. It is not an exhaustive list and there are many more syndromes that can be associated with cardiac lesions. As a general rule, in this situation you should concentrate on diagnosing the cardiac condition (if you recognise the syndrome and know which lesion it is likely to be associated with, this will help you to elicit the signs confidently). The relevance of the syndrome comes when you comment on the dysmorphic features as part of your general observation, and when you seek clarification of the syndromic diagnosis by looking for associated abnormalities in your 'further examination'. For example:

'She is a 6-year-old girl who appears well. On general inspection, she has some features which would be consistent with Turner syndrome. These include … She has features of aortic stenosis which consist of ejection systolic murmur at the upper right sternal edge … Given her other examination findings, I would expect this to be caused by a bicuspid aortic valve. There is no evidence of associated coarctation; specifically she does not have … I would now like to check her blood pressure and plot her growth on a Turner's specific chart.'

Syndrome	*Cardiac lesions*	*Other findings*
Down syndrome	• Up to 50% are quoted to have some congenital heart defect • Ventricular septal defects are the commonest lesions, followed by atrioventricular septal defects • Patent ductus arteriosus and tetralogy of Fallot are less common • All children with Down syndrome should be routinely sent for an echocardiogram • Children with Down syndrome develop pulmonary vascular disease earlier than other children, and hence those with large cardiac defects will develop the Eisenmenger's complex at an earlier stage	• Down facies • Single palmar crease • Sandal gap • Middle ear disease • Hypotonia • Effects on development and growth • Many others
Turner syndrome (XO)	• Bicuspid aortic valve, usually non-stenotic, in one-third of patients • Coarctation of aorta, less common but more serious	• Hypoplastic and hyperconvex nails • Short 4th/5th metacarpals • Widened carrying angle (cubitus valgus) • Low posterior hairline • Prominent ears (look in ears? glue ear) • Neck webbing • High arched palate • Shield chest • Widely spaced nipples • Excessive naevi • 'Also need to examine' – Plot growth – on special Turner syndrome growth chart – Thyroid status (autoimmune hypothyroidism?) – Abdomen (horseshoe kidney?) – Formal audiology – Pubertal staging

Table 8.5: Associations between syndromes and cardiac lesions

Syndrome	*Cardiac lesions*	*Other findings*
Noonan syndrome (phenotypically like Turner, but autosomal dominant)	• Pulmonary stenosis in a usually dysplastic valve, and hence not amenable to treatment by valvuloplasty • Atrial septal defect • Cardiomyopathy	• Short stature • Typical facies – hypertelorism, anti-mongoloid slant, epicanthic folds, ptosis, micrognathia, low-set ears • Wide-spaced nipples, cubitus valgus, short stature and pubertal delay • Cryptorchidism, hernias
Williams syndrome	• Supravalvular aortic stenosis • Branch pulmonary artery stenosis	• Round facies with full cheeks and lips • Stellate pattern in the iris • Strabismus • Supravalvular aortic stenosis • Branch pulmonary artery stenosis • Hypercalcaemia • Mental retardation, friendly personality
diGeorge 22q deletion, also called • CATCH 22 • Velocardio-facial syndrome • Shprintzen syndrome All on a spectrum with subtle differences between each description	• Aortic arch abnormalities • Truncus arteriosus • Pulmonary atresia and ventricular septal defect	May have various combinations of • Structural (cleft)/ functional palatal abnormalities • Nasal speech • Hypotonia • Developmental delay • Hypocalcaemia • Immunodeficiency • Long face • Narrow palpebral fissures • Thin alae nasi • Squared nasal root – bulbous tip • Hypospadias/ cryptorchidism • Long/tapering fingers

Table 8.5: continued

Syndrome	*Cardiac lesions*	*Other findings*
CHARGE association	• Fallot's tetralogy • Ventricular septal defect • Atrial ventricular septal defect • Double outlet right ventricle	• **C**oloboma • **H**eart disease • **A**tresia choanae • **R**etarded growth and development and/or CNS abnormalities • **G**enital anomalies and/or hypogonadism • **E**ar anomalies and/or deafness
VACTERL association	• Fallot's tetralogy • Ventricular septal defect	• **V**ertebral defects • **A**nal atresia • **C**ardiac defects • **T**racheo-oesophageal fistula • **R**enal defects • **L**imb abnormalities This is an updated version of the VATER association
Infant of diabetic mother	• Hypertrophic cardiomyopathy • Ventricular septal defect	• Macrosomia • Sacral agenesis
Trisomy 18 – Edwards syndrome	• Ventricular septal defect • Atrial septal defect • Patent ductus arteriosus • Coarctation of aorta • Bicuspid aortic valve	• Abnormal tone • Microcephaly • Wide fontanelle • Severe growth retardation • Rocker bottom feet • Overlapping fingers • Many other dysmorphic features
Trisomy 13 – Patau syndrome	• Ventricular septal defect • Atrial septal defect • Patent ductus arteriosus • Coarctation • Bicuspid aortic valve	• Scalp defects (aplasia cutis) • Cleft lip and palate • Rocker bottom feet • Polydactyly • Omphalocele • Many other features
Marfan syndrome	• Aortic aneurysm • Aortic regurgitation • Mitral regurgitation	• Tall stature • Hypermobility • Arm span greater than height • High arched palate • Lens abnormalities • Scoliosis

Table 8.5: continued

Syndrome	*Cardiac lesions*	*Other findings*
Ehlers–Danlos syndrome	• Aortic root dilatation	• 'Cigarette paper' scars/ tissue fragility • Easy bruising • Joint hypermobility • Flattened arch of foot
Fetal alcohol syndrome	• Ventricular septal defect • Patent ductus arteriosus • Atrial septal defect • Tetralogy of Fallot	• Absent/flattened philtrum • Developmental delay • Coarse features
LEOPARD syndrome (genetically similar to Noonan syndrome: same site but different mutation)	• Pulmonary stenosis • Long PR interval • Cardiomyopathy	• **L**entigines • **E**lectrocardiographic conduction abnormalities • **O**cular hypertelorism • **P**ulmonary stenosis • **A**bnormalities of genitalia • **R**etardation of growth • **D**eafness Also have characteristic facial features and café-au-lait spots

Table 8.5: continued

Surgery for congenital heart disease

Many of the cardiac cases seen in the exam will have had surgery, often definitive. In some cases it is clear what surgery has been carried out and the underlying diagnosis is clear. For example, a child who is pink with an absent left brachial pulse and a left lateral thoracotomy scar has probably had a coarctation repair. In other cases it is less clear, such as the child with a central sternotomy scar who may have had straightforward surgery, eg ventricular septal defect or the repair of complicated congenital heart disease. It may not be possible to give an underlying diagnosis but a broad knowledge of the principles behind the timing of cardiac surgery, as well as an understanding of what signs to expect at which age for each of the major congenital cardiac lesions, should prevent you from saying things that are clearly impossible.

Procedures can be for repair or palliation.

Repair procedures aim to create a heart that is functionally and anatomically normal. Examples of these include:

- Ventricular septal defect repair
- Atrial septal defect repair
- Tetralogy of Fallot repair (traditionally a full tetralogy of Fallot correction occurs at approx 1 year for size/weight reasons, although definitive repair procedures are now being performed earlier than this because of evidence suggesting improved outcomes)
- Arterial switch procedure for transposition of the great arteries (TGA)

Palliative procedures can be temporary or permanent. Temporary procedures are necessary to prevent or minimise physiological instability pending more definitive surgery. Examples of these could include:

- BT shunt (to allow for pulmonary blood flow, which encourages development of the pulmonary vascular tree, until the child has grown enough to safely have more definitive surgery)
- Pulmonary banding (eg to prevent overloading the pulmonary circulation pending repair of a large ventricular septal defect)
- Atrial septostomy (for example, if there is transposition with inadequate mixing despite a patent duct)

Examples of procedures which are considered permanent palliative procedures include creation of a Fontan circulation.

Timing of surgical intervention

The principles governing the age of surgical correction for individual cardiac defects have already been described in this chapter. The timing and choice of corrective and palliative procedures can vary from unit to unit depending on local expertise. Advances in this field mean that current practice is constantly evolving. Because of this we have tried to avoid being too specific about surgical management. What follows is a description of the general principles behind timing and choice of surgery, so that you can have a system with which to make rational conclusions about the causes of a scar, or the causes of a murmur with no scar, should you encounter this in the exam.

Points to consider in the timing of surgical intervention

Symptoms; for example, poor growth or difficulty feeding due to cardiac failure

Duct-dependent lesions – these may require definitive repair early (in the first week), such as TGA/coarctation, or they may require a palliative procedure with further procedures later

Prevention of long-term problems, eg pulmonary hypertension from left-to-right shunts with increased flow, or ventricular dysfunction in tetralogy of Fallot

The lesion might close spontaneously

Intervention is less risky in larger children

○ Within first week:
  - Duct-dependent lesions require surgery in the first week of life (either repair or palliative depending on condition)
  - Severe obstructive lesions

○ Within first few weeks of life:
  - Some lesions require a fall in pulmonary pressures, either for diagnosis or for physiology which is favourable for surgical repair. Surgery is carried out within the first weeks of life. An example of this could be PA banding, or the creation of a BT shunt
  - Obstructive lesions (eg total anomalous pulmonary venous drainage) could present after the first week of life and be repaired soon after diagnosis

○ Infants:
  - Conditions with a significant left-to-right shunt will require surgery to prevent pulmonary hypertension; this will be done earlier if there is

  concern about faltering growth and cardiac failure, or later if the child is growing well. Occasionally they will have had a PA band to allow for growth before a full repair
  - In general most symptomatic left-to-right shunts will be repaired within the first 6 months of life
  - Lesions with right ventricular outflow tract obstruction (such as Fallot's) may be repaired at this time, avoiding the development of severe right ventricular hypertrophy or cyanotic spells
  - Glenn procedures (cavo-pulmonary shunt) are often performed within the first year of life. The timing could depend on the size of the child, how well they are growing and what their oxygenation is like (falling saturations could indicate they are outgrowing their BT shunt and are ready for the next palliative procedure)
- Older children:
  - Staged repairs – in children who have already had some palliation, for example the conversion of a cavo-pulmonary shunt (superior vena cava to pulmonary artery) to a Fontan circulation (total cavo-pulmonary anastomosis)
  - Surgery to close asymptomatic lesions, to prevent further complications. An example of this could include an atrial septal defect repair, using either a cardiac catheter device or bypass surgery (for larger or unusually shaped lesions)
- Adolescence:
  - Surgery for acquired disease (for example valve replacement following endocarditis)
  - Stent procedures (for example for non-duct-dependent coarctation of the aorta)
  - Repeat procedures, for concerns which have developed following previous cardiac surgery (eg redo pulmonary homograft)

Note that in aortic stenosis and pulmonary stenosis, the timing of operation is more dependent on symptoms and the pressure gradient across the lesion than on age. Ballooning is the favoured treatment. Subaortic stenosis is more likely to cause symptoms because the obstruction is dynamic (worse with exercise).

Long-term post-operative complications following cardiac surgery

- Diaphragmatic palsy
- Arrhythmias (treated with pacemakers/internal defibrillators/Maze procedure)
- Vocal cord palsy
- Protein-losing enteropathy (following Fontan procedure)
- Developmental problems

Chest X-ray findings

The Royal College guidelines clearly state that chest X-ray interpretation would be an appropriate part of the cardiovascular station. You should have a structure for interpreting chest X-rays so that you don't miss anything. The following is an example of a commonly used structure:

1. Check the demographic details. Is this the correct patient? When was the X-ray taken? How old was the patient at the time?
2. Look for the radiographer's markers. Check which side is left and which side is right. Look to see whether it was an AP or a PA film, and whether it was erect or supine.
3. Comment on tubes and lines (eg endotracheal tube, oxygen mask, nasogastric tube, ECG monitoring). The presence of median sternotomy wires should be noted!
4. Look at the lung fields.
5. Comment on the mediastinum. Specifically look at the superior mediastinum and thymus, the cardiac silhouette, and the right and left hilar regions.
6. Look at diaphragms and any abnormalities of the bowel that you can see (eg air under the diaphragm).
7. Look at the bones (ribs, clavicles, shoulders, etc).

If you are looking at an X-ray in the cardiovascular station, it is unlikely that you will have more than 2 or 3 minutes in which to note the key findings. You should think about why the examiner wants you to review the X-ray. There may be clues about the underlying diagnoses, such as a classic X-ray appearance of a particular cardiac lesion (eg a boot-shaped heart). There may be evidence of complications such as congestive cardiac failure or cardiomegaly. If there is an obvious abnormality, you should make your 'structured approach' very brief so that you can skip to the key point. If there are no obvious abnormalities, you will need to be methodical to pick up the extra piece of the puzzle that is being offered to you.

Abnormal cardiac silhouette	**Boot-shaped heart:** tetralogy of Fallot, tricuspid atresia **Egg-shaped heart:** transposition of the great arteries **Cardiomegaly:** seen if there is congestive cardiac failure (eg large VSD, PDA). Can also be seen in TGA **Large globular heart:** Epstein anomaly **Dextrocardia:** look for situs inversus – if the liver is on the same side as the heart, this is often associated with complex cardiac lesions. If the liver is on the left, the heart is often normal
Abnormal shape or position of the liver	Look for associated dextrocardia. The liver could be on the left or in the midline. Remember associated spleen abnormalities and the management of asplenia if asked about the implications of an abnormally sited liver on chest X-ray
Pulmonary oligaemia	Inadequate pulmonary blood flow leads to oligaemic lung fields, and this generally implies a cyanotic lesion. Think of conditions that are associated with severe pulmonary stenosis or atresia
Pulmonary plethora	Think of lesions with a left-to-right shunt (eg VSD/PDA) in an acyanotic baby. If there is cyanosis at birth, think about lesions where there is mixing (eg single ventricle, TGA, truncus arteriosus)

Table 8.6: Cardiac disease on neonatal chest X-rays

Left atrial enlargement	*Seen in mitral valve disorders*
Cardiomegaly	**Acquired cardiomyopathy:** look for associated evidence of cardiac failure **Pericarditis:** 'water-bottle-shaped' heart with increased pulmonary venous markings
Cardiac failure	Upper lobe pulmonary venous congestion Fluid in fissures Pleural effusions Alveolar fluid Horizontal lines in periphery of lower lung fields (Kerley B lines)
Evidence of surgery	Median sternotomy wires Prosthetic valves/grafts Raised hemidiaphragm could indicate intraoperative phrenic nerve damage Chylothorax from damage to thoracic duct

Table 8.7: Acquired abnormalities on chest X-ray

9

ABDOMINAL

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9.1 Introduction

This chapter contains information on examination of the gastrointestinal system.

There is also a detailed review of nutritional assessment and supplementation in this chapter. This can be applied to any pathology, whether gastrointestinal or related to disease in another system. Such knowledge would also stand you in good stead for the history taking/management planning station.

The conditions listed in this chapter are those likely to be seen in the abdomen/'other' stations.

9.2 Clinical examination

Suggested approach for the examination of the gastrointestinal system

Find your own system with which you are comfortable. Always remember to position the child correctly, make them comfortable and maintain good eye contact.

The following is a suggested scheme.

Initial procedure

- Introduce yourself

Inspection – ensure adequate exposure

- Does the child look well?
- Normal or dysmorphic?
- Nasogastric tube, intravenous cannula?
- Well nourished? – comment on nutritional status
- Mention the need to plot on a growth chart (height, weight, OFC if under 2, BMI)
- Look for xanthoma – they are usually on the extensor surfaces, and occasionally around the eyes

Hands

- Clubbing – such a patient in this station may have inflammatory bowel disease, cirrhosis, chronic malabsorption, cystic fibrosis
- Koilonychia – iron-deficiency anaemia
- Palmar erythema
- Pallor

Eyes

- Jaundice
- Anaemia
- Blue sclerae
- Xanthoma

Mouth

- Perioral pigmentation (Peutz–Jeghers)
- Mouth ulceration
- Tongue – stomatitis, large tongue (Down syndrome, Beckwith–Wiedemann syndrome)
- Teeth – should be looked at carefully as they are a good indicator of general nutrition. Dental caries predominant at the back in gastro-oesophageal reflux and at the front with poor diet, eg prolonged bottle feeding with sugary drinks.

Chest

- Spider naevi
- Gynaecomastia

Abdomen

- Scars (laparoscope, groin, loin)
- Umbilical hernia
- Abnormal vessels – caput medusae (drain from the umbilicus)
- Distension
- Superficial palpation – all over once
- Deep palpation – all over once
- Liver
- Spleen
- Kidneys

Further examination

This should be done as appropriate and depends on the previous findings. For example, it is not necessary to look for shifting dullness if the abdomen is not distended:

- Percussion
- Bowel sounds
- Abdominal distension – inspect from the side of the bed/couch, stand child up and look at buttocks, as they may also be wasted, suggesting malabsorption
- Ascites – fluid thrill, shifting dullness (only if distended)
- Can I see the back?
- Can I see the genitalia?
- Hernia orifices

Notes on abdominal scars

An abdominal scar(s) would not be an uncommon way to begin a short case. Such scars could be from open or laparoscopic surgery. Older children may have had open procedures when younger that would be performed laparoscopically these days, so you need to be familiar with scars from both types of surgery. You will need to understand the possible procedures that may have resulted in that scar and to be able to talk about them.

Laparoscopy scars

These are becoming more frequently seen as laparoscopic GI and renal surgery is now common. Scar sites are usually small and may be multiple. A shortlist of the commonest laparoscopic procedures would include:

- ○ Inguinal hernia repair
- ○ Pyloromyotomy for pyloric stenosis
- ○ Appendicectomy
- ○ Cholecystectomy
- ○ Fundoplication
- ○ Total or partial nephrectomy

Laparotomy scar

The finding of a laparotomy scar may be secondary to a wide variety of intra-abdominal pathology, and it is important to look for surrounding clues (particularly other scars) that might lead you to the diagnosis. Context is all-important:

- ○ **Down syndrome baby** with a laparotomy incision: look for the site of a colostomy on the abdomen, and think of **Hirschsprung's disease**; alternatively, if there is a laparotomy scar and no colostomy scar there may have been a duodenal or a small or large intestinal **atresia**
- ○ Are there any signs of **cystic fibrosis**, and could this be a **meconium ileus** repair?
- ○ Could this be an **ex-premature infant** who had **necrotising enterocolitis** during the neonatal period?
- ○ Is there an **associated thoracolumbar incision** (may be laparoscopy scars instead) suggesting a **renal transplant**? Feel for mass in right iliac fossa as this is the site of implantation of the donor kidney
- ○ **Other possibilities:**
  - Kasai procedure for biliary atresia
  - Malrotation
  - Intussusception
  - Tumour removal

Other scars – by site

- **Below right subcostal margin – Kocher's incision.** Allows access to the liver and the biliary tree and is seen after cholecystectomy and gallstone removal. Patients with sickle cell disease or hereditary spherocytosis may have had such surgery in the past. It is now more commonly performed laparoscopically.
- **Transverse upper abdominal incision** – repair of congenital diaphragmatic hernia. These are more common on the left than the right. This procedure can be performed laparoscopically in some cases.
- **Upper abdominal midline incision** – Nissen's fundoplication. Does this child have neurodevelopmental problems? Is there an associated gastrostomy scar? This procedure is now mostly performed laparoscopically.
- **Small right upper transverse ('rectus cutting') incision** is the site of Ramstedt's pyloromyotomy, more common in boys. Can be performed laparoscopically.
- **Right iliac fossa scar** – open or laparoscopic appendectomy.
- **Lateral thoracolumbar incision** – nephrectomy. Commonly performed in children with a cystic dysplastic kidney to prevent infection in the redundant kidney. Can be performed laparoscopically in suitable cases.
- **Sub-umbilical/umbilical scar** – suggests gastroschisis or exomphalos, again think of associated conditions. The absence of a normal 'belly button' is an important clue. Alternatively could be an umbilical hernia repair.
- **Groin scars** suggest inguinal hernia repair, more common in ex-premature male infants. Can be performed laparoscopically.

Examination of the liver, spleen and kidneys

- Important to do correctly
- Liver/spleen – start in right iliac fossa
- If organomegaly found, confirm with percussion – measure if appropriate

Liver

- Edge – regular or irregular
- Surface – smooth or nodular
- Texture – firm or hard
- Tenderness
- Is there a rub?
- Is there a bruit?

Spleen

- As above
- Is there associated hepatomegaly/ascites?
- Other associations (eg jaundice and hereditary spherocytosis)

Common exam questions

- ○ How do you differentiate between a liver, spleen and kidney?
- ○ Why is it a liver?
- ○ Why is it a spleen?

These are straightforward but commonly asked questions that are often answered badly. If you find organomegaly it is important that you assess its character carefully and make it clear to the examiner using the criteria below that you are demonstrating a liver, spleen or kidney.

Liver

- Right hypochondrium
- Cannot get above it
- Moves with respiration
- Dull to percussion

Spleen

- Left hypochondrium
- Cannot get above it
- Moves with respiration
- Dull to percussion
- Has a notch

Kidney

- Can get above it
- Doesn't move with respiration
- Resonant
- Ballotable

Tip +

Whenever you are practising, get into the habit of justifying your findings in such a manner

9.3 Nutrition

Assessment of nutritional status

The assessment of nutritional status is very important and knowledge can be tested in different sections of the exam. The information in this section could help you in more than one station! Firstly, the general case and appropriate comments are considered, then specific scenarios with notes on how to deal with them are provided.

Instructions to candidate

Look at this 10-year-old boy and comment on his nutritional status.

Case

> *John is comfortable at rest. He looks thin. I would like to plot him on a growth chart. His muscle bulk is poor with a reduced mid-arm circumference and his subcutaneous fat mass is poor with a reduced skinfold thickness. I note he has a gastrostomy tube in situ. I would like to proceed with a full examination. He looks pale ...*

Notes on nutritional assessment

This is an essential part of the general examination of a patient and applicable to children with pathology from the gastrointestinal or other systems. Commenting on nutritional status is all part of making a general comment about a patient. If in any doubt about nutritional status it is sensible to suggest to the examiner that you would plot the child onto a height and weight chart. Even if your performance of a measurement, eg skinfold thickness, may not be tested, you will still be expected to ask to measure/plot it. If the nutrition looks poor then the following scheme may be helpful.

Scheme

- ❍ Look for and comment on obvious pathology, eg cystic fibrosis.
- ❍ Presence of nasogastric tube, gastrostomy
- ❍ Nails (koilonychia)
- ❍ Pallor – hands, mucous membranes, skin
- ❍ Hair – thinning
- ❍ Pubertal status, if older
- ❍ Carefully plot height, weight and head circumference:

  - You may be asked how you would derive body mass index (BMI), which is weight in kg divided by the square of height in metres
  - Be prepared to comment on **weight for height**, which is a comparison of the patient's weight with the 50th centile weight of a child of the same height. It is expressed as a percentage of the expected weight
- ❍ Triceps and subscapular skinfold thickness, measured with callipers, to give an indication of the subcutaneous fat mass
- ❍ Mid-upper arm circumference impression of muscle bulk

Notes on malnutrition

It is useful to give some thought to potential mechanisms as poor nutrition is frequently multifactorial. Cystic fibrosis provides the best example and is commonly seen in the exam. Others include congenital heart disease, bronchopulmonary dysplasia, severe neurodevelopmental disorders and chronic inflammatory bowel disease.

The aetiology of malnutrition is often complex and various factors play a role. For example, in cystic fibrosis:

- ❍ Malabsorption causing increased losses
- ❍ Increased energy needs:
  - Chronic cough
  - Dyspnoea
  - Recurrent infection
  - Inflammation
- ❍ Reduced intake:
  - Anorexia
  - Vomiting
  - Psychological problems resulting in disordered eating

All these together result in an energy deficit. All factors need to be taken into account when nutritional supplementation is considered.

Nutritional supplementation/concentrating feeds

Nutritional supplementation should be done with the help of a paediatric-trained dietician. Try to attend a dietetic clinic to observe the dietetic management at first hand.

Basic information – normal weight gain

- ❍ Normal birth weight for term infants in the UK is 3.3–3.5 kg for both sexes
- ❍ Loss of weight occurs in the first 5–7 days, then birth weight should be regained within the next 14 days

- Thereafter, average weight gain is:
  - 200 g/week for first 3 months
  - 150 g/week for second 3 months
  - 85–100 g/week for third 3 months
  - 50–75 g/week for fourth 3 months

Supplementation

Nutritional supplements (see Box) are generally required for those infants with faltering growth on normal feeds, or who are fluid restricted. Care needs to be taken not to present too high an osmotic load as diarrhoea may result.

Carbohydrate supplements

- Glucose polymers are used: examples include Caloreen®, Maxijul®, Polycal® (all hydrated cornstarch)

Fat supplements

- Usually fat emulsions, examples include:
  - Calogen® – arachis oil (not if peanut allergic as it is a crude oil and so may have traces of protein)
  - Liquigen® – medium-chain triglycerides (MCT) oil

Combined carbohydrate and fat supplements

- Duocal® – cornstarch, maize oil and coconut oil

Protein supplements

- Whole protein, peptides or amino acids
- Rarely required without an accompanying increase in other energy sources
- Examples include Maxipro HBV®, which is whey protein

Concentrating infant feeds

Concentrating infant feeds increases the density of energy and protein, and has the advantage of providing this without the need to add a series of supplements. If feeds are concentrated then this also concentrates up electrolyte, vitamin and mineral replacement, which may be a problem.

- Most normal baby milks are made up to 13%, providing 65 kcal per 100 ml and a protein content of 1.5 g/100 ml. Making a feed up to 15% provides 75 kcal/100 ml and 1.7 g/100 ml of protein
- Carbohydrate and fat can still be added if required

There are multiple commercially available high-calorie-density milks that have the appropriate electrolyte, mineral and vitamin contents, eg Infantrini® (100 kcal/100 ml), Nutrini® (100 kcal/ml), Nutritini Energy® (150 kcal/ml) (Nutricia®).

Malnutrition – cases

Specific scenarios arise within the different systems and need to be considered.

Note that such questions could also be asked at the end of the history taking/ management planning station when you are left with the examiner.

Case one

This 6-month-old infant has congenital heart disease with faltering growth – comment on his nutritional status. What nutritional supplementation would you recommend?

In this infant the poor nutritional state will be a consequence of increased metabolic demands (increased work of breathing, periods of hypoxia) and poor intake secondary to breathlessness. Liaison with a dietician and supplementation to increase the calorie density of feeds would be appropriate, together with consideration of other methods of administration (eg nasogastric tube).

Case two

This 6-month-old infant has chronic lung disease and has had severe faltering growth. Comment on possible causes.

In this infant the poor nutritional state will be as a consequence of increased metabolic demands (for example work of breathing causing increased energy requirements) and poor intake secondary to breathlessness. In addition, other factors may be relevant such as gastro-oesophageal reflux. Supplementation to increase calorie density would be appropriate. This may have to be via a nasogastric tube. Gastrostomy can be considered. Investigation for gastro-oesophageal reflux may be appropriate, as treatment in tandem with nutritional management is likely to result in better weight gain.

Case three

This 13-year-old boy has cerebral palsy with bulbar involvement. Comment on his nutritional status. What strategies can be used to improve it?

This child's principal problem will be with intake, either because of reflux or secondary to the bulbar problems, or both. In addition to being given nutritional supplements, this child would benefit from help with feeding. I would refer him to the speech and language therapist and occupational therapist. Consideration also needs to be given to nasogastric tube or gastrostomy tube feeding if appropriate. In some instances where reflux is a particular problem, a fundoplication or consideration of gastrojejunal feeding will be required.

Case four

This boy has cystic fibrosis and is malnourished. Comment on the cause. What can be done to help?

His energy requirements will be high due to chronic cough and airway inflammation, and calorie supplementation with energy-dense supplements will be needed. Children with cystic fibrosis often have a poor appetite due to chronic malaise, and therefore poor intake could also be part of the problem. The additional factor is malabsorption, for which pancreatic and vitamin supplementation will be required.

Emerging CF-related diabetes should also be excluded as a factor contributing to weight loss.

This child will need regular dietetic assessment and may require either a nasogastric tube or a gastrostomy to aid administration of his feeds and supplements.

Gastrostomy tube feeding

It is essential to have seen and to recognise a gastrostomy tube. They are generally inserted endoscopically (percutaneous endoscopic gastrostomy). Complications are rare.

Indications

- In chronic disease (eg cystic fibrosis, bronchopulmonary dysplasia) where there is nutritional impairment despite attempts by the multidisciplinary team to optimise intake
- For intensive nutritional therapy in Crohn's disease
- Because of difficulties with feeding (eg cerebral palsy, particularly with an associated bulbar palsy, and other neurodevelopmental disorders)
- In severe gastro-oesophageal reflux with a fundoplication
- Children dependent upon nasogastric feeding for any other reason

It is also important to consider that children may have gastrojejunal tubes. These are used for feeding children (particularly those with neurodisability) in whom gastrostomy tube feeds are poorly tolerated and fundoplication is not appropriate.

Obesity

This is defined as a body mass index (BMI) above the 98th centile for age and sex. There is an increase in childhood obesity in many parts of the world, with the UK being no exception. The causes are nearly always due to excess energy intake with reduced expenditure. Childhood obesity is a strong predictor for obesity in adulthood.

Current guidelines on obesity from the National Institute for Health and Care Excellence (NICE) (www.nice.org.uk/guidance/cg43) recommend consideration of tailored clinical interventions for children with a BMI at or above the 91st centile. Children with a BMI at or above the 98th centile should undergo a co-morbidity assessment.

It is unlikely that you will be asked to examine a child in the clinical section who is obese due to excess food intake, but remember that there are other causes of obesity (see Box).

It may also be that weight is relevant in one of the other cases you see.

In your daily practice you should be aware that obesity which persists into adulthood is a risk factor for type 2 diabetes, hypertension, ischaemic heart disease, stroke and certain cancers. Obese children are at major risk of becoming obese adults.

Remember that children with an endocrine or genetic condition are usually short as well as overweight and may have dysmorphic features. Children with physiological obesity are usually tall.

Other Causes Of Obesity (less than 5% of cases)

- Chromosomal/genetic:
  - Down syndrome
  - Kleinfelter syndrome
  - Prader–Willi syndrome
  - Lawrence–Moon–Biedl syndrome
- Endocrine:
  - Growth hormone deficiency
  - Hypothyroidism
  - Cushing syndrome or prolonged treatment with corticosteroids
  - Polycystic ovarian syndrome
- Oncological:
  - Pituitary tumour

Physiological obesity/metabolic syndrome

Problems that obese children face (or that they are likely to face as they get older) include hypertension, non-alcoholic steatohepatitis, gallstones, hypercholesterolaemia, insulin resistance (associated with acanthosis nigricans), type 2 diabetes and obstructive sleep apnoea. Many of these young people are bullied at school, and depression is increasingly being recognised in this group. Remember that girls who are adolescent and obese are at increased risk of polycystic ovary syndrome.

If you are asked what investigations to perform to look for co-morbidities or causes of a BMI greater than the 98th centile, remember that as a baseline NICE recommends that you check the following:

- Thyroid function
- Fasting glucose
- Lipid profile
- Insulin levels
- Liver function tests

Management of functional obesity

- Prevention – public health campaigns, eg healthier school dinners, free fruit, banning drinks machines in schools, reduction in TV advertising of 'junk foods'.
- Weight reduction programmes – these are most effective when the whole family is on board, the multidisciplinary team is involved and goals are realistic. Such programmes involve changing behaviours. Healthy eating,

portion control and 'prescribed' exercise are all components of such programmes. A reduction in sedentary behaviour is also helpful (eg limiting time watching TV/on the computer to less than 2 hours a day). NICE advocates that 'Dietary approaches alone are not recommended'.

- Drug treatment with Orlistat (a lipase inhibitor) for over 12-year-olds is an option in severely affected children, with significant co-morbidities, who have been assessed and felt suitable candidates by a specialist multidisciplinary team.

9.4 Cases and conditions

Cystic fibrosis

Case one: 12-year-old boy

> *Peter is a thin-looking boy and I would like to plot him on a growth chart. He has reduced subcutaneous fat and muscle bulk. There is a gastrostomy tube in situ. He is clubbed. I note on further inspection that he is tachypnoeic and there are Harrison's sulci present. Examination of his abdomen reveals hepatomegaly which is firm in consistency. There is splenomegaly and caput medusae ...*
>
> *I suspect he has cystic fibrosis complicated by liver disease – presumably cirrhosis associated with portal hypertension ...*

Case two: 10-year-old girl

> *Sarah is a well-looking White girl. I would like to plot her growth parameters. She is clubbed. She has a hyperexpanded chest. On examination of the abdomen there is a transverse incision. She also has a palpable mass in the right iliac fossa.*
>
> *I wonder if she has cystic fibrosis and distal intestinal obstruction syndrome ...*

Gastrointestinal manifestations of cystic fibrosis

More detailed notes can be found in Chapter 10.

Pancreatic

- Insufficiency occurs in around 85% of cases, and presents with steatorrhoea and poor weight gain
- Pancreatitis
- Abnormal glucose tolerance in up to 10% by the second decade
- Insulin-dependent diabetes mellitus in 7% of patients; remember that diabetic ketoacidosis is rare in this group

Intestinal

- ○ Meconium ileus
- ○ Atresias
- ○ Rectal prolapse
- ○ Distal intestinal obstruction syndrome (DIOS)
- ○ Chronic strictures secondary to high-dose pancreatic supplementation

Hepatobiliary

- ○ Cholestasis in infancy – leads to obstructive jaundice
- ○ Fatty liver
- ○ Focal biliary fibrosis
- ○ Multilobular cirrhosis
- ○ Abnormalities of the gall bladder
- ○ Cholelithiasis
- ○ Obstruction of the common bile duct

As some patients develop jaundice, ascites and portal hypertension secondary to hepatobiliary pathology, you must always comment on the presence or absence of these features when asked to present your GI findings in a child with cystic fibrosis.

Crohn's disease

This may be seen in more than one section of the exam and therefore a good knowledge base of the various aspects is required. The clinical signs are not particularly specific but an illustrative case is included.

Case

> *This 14-year-old boy looks pale and thin. I suspect he is short for his age and I would like to plot him on a growth chart. His muscle bulk and subcutaneous fat mass are poor. I would like to look in his mouth for lip/gum swelling and aphthous ulceration. There is mild right iliac fossa tenderness but no other abnormalities on abdominal examination.*
>
> *I would like to check his pubertal status and examine his peri-anal region for skin tags, fissures and fistulae.*
>
> *Finally, I would like to look for any extra-intestinal manifestations by examining his skin and joints.*

There may also be:

- Nasogastric tube or gastrostomy
- Signs of steroid toxicity
- Scars from previous surgery

Notes on Crohn's disease

Crohn's disease is an idiopathic chronic inflammatory disorder of bowel involving any region from mouth to anus, with many cases involving the terminal ileum. The inflammation is transmural (so effectively narrowing the lumen) with skip lesions. Affected mucosa is granulomatous and interspersed with deep ulcers, giving a cobblestone appearance.

There has been an increase in the incidence of inflammatory bowel disease since the late 1990s, and in children under 16 years of age it now stands at just over 5 per 100 000.

Symptoms and signs

- The commonest presenting symptoms are abdominal pain, diarrhoea and weight loss
- Systemic upset, lethargy and fever can occur
- Pallor secondary to anaemia
- Extra-intestinal manifestations are worth looking for in a known case:
  - Eyes – uveitis or episcleritis
  - Joints – peripheral arthritis
  - Skin – erythema nodosum, pyoderma gangrenosum, erythema multiforme
- Growth failure with delayed bone maturation and delayed sexual development is common. The aetiology of this is complex and includes poor nutritional state, inflammatory disease activity, endocrine disturbance and the side-effect of corticosteroids when used.

Diagnosis

This is made on the basis of the clinical history, raised inflammatory indices (CRP, ESR, stool calprotectin) and diagnostic tests. The latter include ileo-colonoscopy and upper GI endoscopy (with biopsies), and radiological tests. Imaging studies (ultrasound, MRI with contrast in particular) can help to determine the extent of small bowel disease and identify strictures.

MRI is also a useful tool in the diagnostic work-up of a patient with peri-anal disease.

Treatment

The disease usually runs a chronic relapsing course. The aim of treatment is to induce a disease remission and facilitate normal growth and development while minimising treatment toxicity:

- Corticosteroids for initial presentation or relapses, particularly if systemic upset is present. Once remission is achieved these can be tapered down. Dependence or resistance can occur and additional immunosuppression with drugs such as azathioprine (steroid sparing) may be necessary.
- Exclusive enteral nutrition is another option, with an elemental or polymeric diet lasting for up to 8 weeks followed by a period of controlled food reintroduction. This induces remission in up to 80% of patients. Repeat courses may be needed for relapses.
- Anti-inflammatories – oral 5-aminosalicylic acid (5-ASA) derivatives can be useful in some cases, especially if still deteriorating despite steroids, or not tolerating them. Rectal disease can be treated with 5-ASA enemas.
- Anti-tumour necrosis factor (TNF) monoclonal antibody therapy such as infliximab or adalimumab are increasingly used in severe/refractory cases.
- Antibiotics for abscesses and fistulae – ciprofloxacin and metronidazole.
- Surgery – indicated if there are persistent symptoms despite optimal medical therapy, intolerable side-effects of medical therapy, abscesses that are unresponsive to conservative treatment or life-threatening toxic megacolon. The need for surgery increases with age. Relapse following surgical resection is common.

Coeliac disease

This may be seen in the abdomen station or the history taking/management planning station (see Chapter 3).

Case

Examine this child and suggest a diagnosis.

Clinical features:

- May be relatively short
- Pale
- Thin with reduced subcutaneous fat, poor muscle bulk and buttock wasting
- Abdominal distension

This 2-year-old girl looks pale and is thin. I would like to plot her on a growth chart. Her muscle bulk and subcutaneous fat mass are reduced. Abdominal examination is unremarkable apart from marked distension. On standing, her distension increases and I can see buttock wasting. I suspect she has a malabsorption syndrome.

Coeliac disease would be the most likely diagnosis.

Supplementary questions (could also apply to history taking/management planning station)

- ○ What are the investigations?
- ○ What is the management?
- ○ What are the indications for a gluten challenge?

Notes on coeliac disease

Coeliac disease is an immune-mediated gluten-sensitive enteropathy, and is considered a systemic disorder. There is genetic susceptibility (HLA-DQ2 or DQ8 haplotypes) with an increased prevalence in first-degree relatives. The intolerance is to gluten, which is present in wheat, rye and barley. Oats are usually cross-contaminated during the milling process involved in their production.

The current prevalence in Western Europe is in the range 0.3%–2%, with UK prevalence thought to be around 1%. Coeliac disease is relatively more common in Western Ireland. There are certain conditions that carry a higher risk of the child also having coeliac disease. These include Turner syndrome, Down syndrome, autoimmune liver/thyroid disease, and type 1 diabetes. Indeed, children with diabetes are screened for coeliac disease at diagnosis and 3-yearly thereafter.

Symptoms and signs

- ○ Symptoms of coeliac disease usually present after 6 months of age (ie after gluten has been introduced into the diet) with chronic diarrhoea and poor weight gain, with abdominal distension.
- ○ Children more often present with fairly non-specific gastrointestinal symptoms, including chronic abdominal pains, nausea, constipation, anorexia, flatulence.

Other features include:

- ○ Anorexia
- ○ Lethargy.

- Irritability – patients are characteristically very miserable
- Pallor (iron-deficiency anaemia)
- Dermatitis herpetiformis is occasionally seen and is an intensely itchy, blistering skin rash
- Recurrent apthous stomatitis
- Rickets, osteomalacia

Diagnosis and treatment

NICE has published guidelines (www.nice.org.uk/cg86) on the diagnosis and treatment of coeliac disease. The European Society of Paediatric Gastroenterology, Hepatology and Nutrition (ESPGHAN) guidelines are also used.

In terms of investigations, children are classified according to whether they are symptomatic or asymptomatic. Note that false-negative tests can occur if children have been taken off gluten before testing; most guidelines stipulate that the minimum gluten intake required to avoid false-negative investigation results is about 2 grams a day, for at least 6 weeks. If in doubt, a dietician may need to be consulted.

If a child is symptomatic, recommended first-line test is a tissue transglutaminase (TTG-IgA) test, coupled with an IgA level. False-negatives are seen in children who are IgA deficient, and around 10% of children with coeliac disease have a selective IgA deficiency. IgG antibodies can be used in this group.

If the TTG comes back at over ten times the upper limit of normal, this is highly suggestive of villous atrophy. A biopsy can be avoided (after discussion with a paediatric gastroenterologist) if a repeat TTG is similar, anti-endomysial antibodies are positive and the patient is HLA-DQ2/8 positive on genetic testing. If the first TTG is high but less than ten times upper limit of normal, one would normally proceed to duodenal biopsy.

Asymptomatic children with a raised TTG can be tested for presence of HLA-DQ2/8 haplotype. If positive, a biopsy is recommended, no matter how high the original TTG. If negative, coeliac disease is highly unlikely.

In children who do undergo biopsy, diagnosis is confirmed by the demonstration of characteristic histological features of the duodenum or jejunum. These features are sub-total villous atrophy, crypt hyperplasia, intra-epithelial lymphocytes and a lamina propria plasma cell infiltrate.

Treatment is with a gluten-free diet for life, which should result in remission of symptoms within a few weeks. If it does not, the diagnosis should be reviewed. Follow-up with a paediatric gastroenterologist (or a paediatrician with a special interest) along with a dietician is very important. The gluten-free diet itself has no long-term complications. If this diet is not adhered to, there is a long-term risk of small bowel lymphoma. Much commoner complications of non-compliance are ongoing short stature, osteoporosis, iron-deficiency anaemia and delayed puberty These are in themselves presentations of undiagnosed coeliac disease.

You may be asked to name the indications for a gluten challenge; this test is not routinely recommended and main indication is initial diagnostic uncertainty. This scenario is rarer than it used to be now there are very helpful diagnostic guidelines.

Differential diagnosis of the 'malabsorptive' picture (= malabsorption syndrome)

- ○ Coeliac disease
- ○ Cows' milk protein allergy (non-IgE-mediated form)
- ○ Cystic fibrosis
- ○ Immunodeficiency
- ○ Others = many, eg infection, giardiasis, soya intolerance, autoimmune disease, etc

Constipation

This is the commonest cause of an abdominal mass. It has been reported by candidates to be a common short case; children may be recruited from the ward on exam day if there is a shortfall in cases.

Case

This 9-year-old girl looks well. I would like to plot her on a growth chart ... proceed through examination ... She has a full abdomen with a palpable mass in the left lower quadrant. This mass is hard and indents. I suspect it is a faecal mass and that she has chronic constipation. I would like to inspect her peri-anal region as she may have overflow soiling and it is essential to exclude local pathology such as a fissure.

What else would you examine ...

Her lower back appears normal with no signs of spina bifida ... she has normal tone, power, reflexes and sensation in her lower limbs ... I would also like to test saddle sensation.

Notes on constipation

Most children with constipation do not have an underlying cause and their constipation is idiopathic. If associated with a megarectum, then soiling is common. This should be distinguished from encopresis, a primarily psychological problem in which stool is passed at inappropriate times and in inappropriate places with no underlying constipation. True encopresis in the absence of constipation is rare.

Underlying causes that need to be considered include:

- ○ Hirschsprung's disease – this is very rare in children who have at some stage of their life had a normal bowel habit and usually presents in the neonatal period
- ○ Thyroid disease (comment on presence or absence of goitre)
- ○ Meconium ileus equivalent
- ○ Spina bifida

The management of chronic idiopathic constipation is multidisciplinary, and guidelines have been published by NICE (www.nice.org.uk/guidance/cg99). Many factors may need to be addressed, including local pathology (eg anal fissure), poor diet, low fluid intake, previous difficult toileting experiences and psychosocial problems. Drugs that are used to treat the condition include osmotic (eg macrogols, lactulose) and stimulant (eg senna) laxatives. Please refer to Chapter 3, Section 3.6, for an in-depth consideration of history taking and management planning for this condition.

Umbilical hernia

A common condition, which presents in infancy. Unlike inguinal hernias of infancy, it almost never obstructs in children. Usually resolves by age 2 years. If it has not resolved by then, it is unlikely to and therefore surgery should be considered at around 4 or 5 years of age.

Remember the associated conditions:

- ○ Hypothyroidism
- ○ Storage disorders, eg mucopolysaccharidoses
- ○ Beckwith–Wiedemann syndrome
- ○ Trisomy 18
- ○ Trisomy 13

Ascites

What is ascites?

How do you examine for it and what are the potential causes?

Clinical features (not obvious clinically in 50%)

- ○ Bulging of flanks
- ○ Protrusion of umbilicus
- ○ Scrotal swelling
- ○ Fluid thrill
- ○ Shifting dullness

Differential diagnosis

Wide – four main groups of pathologies:

- ○ Cirrhosis
- ○ Congestive cardiac failure
- ○ Nephrotic syndrome
- ○ Protein-losing enteropathy (rare)

Hepatomegaly and splenomegaly

Hepatomegaly, splenomegaly and hepatosplenomegaly may well be encountered in the abdominal station. It is important to have a wide differential diagnosis, but try to focus on the most likely causes in the patient you have seen.

Causes of splenomegaly that are most relevant to this station are covered in this chapter, but some other causes of splenomegaly are discussed in the section on haematology in Chapter 12. We have tried to avoid duplication where possible.

Hepatomegaly

- ○ Glycogen and other storage disorders
- ○ Right-sided heart failure

Splenomegaly

- ○ Infection – glandular fever
- ○ Portal hypertension
- ○ Hereditary spherocytosis
- ○ Sickle cell disease
- ○ Idiopathic thrombocytopenic purpura
- ○ Myeloproliferative disease

Hepatosplenomegaly

- ○ Congenital hepatic fibrosis
- ○ Cystic fibrosis
- ○ Thalassaemia
- ○ Mucopolysaccharidoses

Case

Examine this child's gastrointestinal system. Talk me through your examination.

This is a 10-year-old girl. She looks well. She appears relatively small. I would like to plot her on a height and weight chart.

(Proceed to examine – lift hand)

There is no evidence of clubbing or palmar erythema.

(Her facies are a little immature – may or may not notice; assess conjunctiva)

There is no pallor or jaundice. Her mouth looks clear with no aphthous ulceration or stomatitis. There are no spider naevi on the chest. Her abdomen is full. There are no scars or abnormal vessels.

(Palpate – superficial then deep palpation – should at this stage be able to feel the large liver, then assess formally)

She has a large liver palpable in the right hypochondrium. The liver is hard, non-tender and measures 6 cm below the costal margin.

(Percuss to confirm upper border; feel for spleen)

There is no associated splenomegaly. Kidneys are impalpable.

(Occasionally may be impalpable). (If clear about findings can stop there)

In summary, this is a well 10-year-old girl. She has massive hepatomegaly but no other signs of liver disease. I suspect she may have glycogen storage disease type one

Notes on hepatomegaly

You may be asked for a full differential diagnosis of hepatomegaly, in which case you will need to have one available. We have included one below based on different categories – infection, infiltration, idiopathic, storage disorder, obstruction, miscellaneous. Avoid mentioning rarities and give just one or two examples in each category.

You don't want to be mentioning some rare differential you don't know much about and then be asked about it by the examiner – black hole! **The most likely scenario is that you will be asked for a diagnosis that would be appropriate in the child you have seen.** For example, you would not expect to be taken to see a child who is jaundiced with a large tender liver and infective hepatitis.

Equally, you would not expect to be asked to palpate the liver of a child with extensive metastases. You are more likely to see a stable and well child with a glycogen storage disease or other storage disorder, or a child with portal hypertension or a stable child post-Kasai procedure for biliary atresia. Such patients will usually be able to tolerate repeated, similar examinations a few times in a row if required. This means you need to ensure you give an appropriate and relevant answer to the questions asked and remember that many causes of organomegaly in the abdomen are not likely to be used as short cases for the exam.

There may be no physical signs in a child with liver disease such as chronic hepatitis B infection.

Differential diagnosis of hepatomegaly

- Infection:
  - Hepatitis A
  - Hepatitis B
  - Epstein–Barr virus (EBV) infection
- Infiltration:
  - Primary liver tumours
  - Secondary tumour, eg lymphoma, leukaemia, neuroblastoma
- Idiopathic
- Storage disorders:
  - Fat:
    - Cystic fibrosis
    - Obesity
    - Malnutrition
    - Total parenteral nutrition (TPN)
  - Lipid:
    - Gaucher's disease
  - Sphingomyelin – cholesterol:
    - Niemann–Pick's disease
  - Glycogen:
    - Glycogen storage disease – see notes below
- Obstructive:
  - Hepatic vein thrombosis
  - Congestive cardiac failure
- Miscellaneous:
  - Wilson's disease
  - Alpha 1 antitrypsin deficiency
  - Congenital hepatic fibrosis

Very large liver

You may examine a patient with a huge liver – remember to start palpating low down in the right iliac fossa!

- ❍ Storage disorder
- ❍ Reticuloendothelial disease, eg leukaemia
- ❍ Gross fatty change
- ❍ Malignancy
- ❍ Congestive cardiac failure

Glycogen storage disease

Notes on glycogen storage disease type I – von Gierke's disease

An example of a very rare disorder with disproportionately high representation in the exam. You may not have encountered such a case so far in your career, so you will need to fall back on your notes!

This is an autosomal-recessive disorder and is the most common glycogen storage disease seen. It is due to reduced glucose-6-phosphatase activity and usually presents within the first 6 months with **hypoglycaemia** or **asymptomatic hepatomegaly**. Diagnosis used to be from liver biopsy with estimation of hepatic glucose-6-phosphatase activity, but genotyping can now be used.

Clinical findings

- ❍ Often short
- ❍ Doll-like facies (particular pattern of fat deposition)
- ❍ Large liver – due to the accumulation of glycogen and fat
- ❍ Large kidney – occasionally

Main problems

- ❍ Can present as fasting hypoglycaemia with lactic acidosis, which can cause convulsions. There is no response to glucagon
- ❍ Patients need frequent or continuous feeds (via nasogastric tube), which is even more important when unwell. Intravenous dextrose may of course be required
- ❍ For older children cornstarch (a slow-release carbohydrate) is used. Overnight feeds often still needed
- ❍ Long-term risk of renal calculi and hepatoma; screened by ultrasound
- ❍ Platelet dysfunction
- ❍ Puberty is often delayed

These children are normally under a metabolic team and should have an emergency feeding protocol.

Notes on other storage diseases that cause hepatomegaly

Glycogen storage disease type III (Cori's disease)

This is a debranching enzyme deficiency of liver and muscle. Important clinical features are:

- Massive hepatomegaly
- Myopathy
- Developmental delay

Patients often develop cardiomyopathy in their late teens or adulthood. Liver cirrhosis is also a risk.

Glycogen storage disease type IV (Andersen's disease)

A rapidly progressive hepatic disease with hepatomegaly, ascites and portal hypertension leading to cirrhosis.

Gaucher's disease

- Autosomal recessive with different forms – infant, juvenile and adult
- Deficiency in glucosylceramide beta-glucosidase (glucocerebrosidase) resulting in lipid accumulation in reticuloendothelial cells
- Can progress to portal hypertension and cirrhosis

Splenomegaly/portal hypertension

Case

Examine this child and suggest a likely diagnosis.

Callum is a 10-year-old boy who appears well-grown. He is pale. Caput medusae are visible. His abdomen is full but not distended and he has a large spleen extending to 6 cm below the costal margin. Ascites is also present. No other signs.

The likely diagnosis is portal hypertension secondary to pre-hepatic portal vein obstruction.

What are the complications of portal hypertension?

Notes on splenomegaly

The aetiology of splenomegaly can be split into the following categories:

- Hyperplasia of the reticuloendothelial system
- Excessive antigenic stimulation – infection
- Disorders of immunoregulation – autoimmune disease
- Excessive destruction of blood cells – haemolytic anaemia
- Neoplasia
- Disorders of splenic flow – portal hypertension
- Infiltration – metabolic lesions
- Extramedullary haemopoiesis

Differential diagnosis of splenomegaly

Infection (bacterial, viral or protozoal)

- Septicaemia
- Infectious mononucleosis
- Malaria

Autoimmune disease

- Juvenile idiopathic arthritis (JIA)
- Systemic lupus erythematosus (SLE)

Haemolytic disorders

- Hereditary spherocytosis
- Sickle cell anaemia (early)

Neoplastic

- Acute leukaemia
- Hodgkin's disease

Disordered splenic flow

- Portal hypertension
- Cirrhosis
- Cardiac failure

Infiltration

- Gaucher's disease

Extramedullary haemopoiesis

- Thalassaemia

Notes on portal hypertension

The cardinal feature of portal hypertension is splenomegaly. **Portal hypertension does not necessarily imply liver disease:**

- If there are no clinical or biochemical features of liver disease and the liver is not enlarged, portal vein obstruction is the most likely diagnosis
- If the liver is large and there are no stigmata of cirrhosis then congenital hepatic fibrosis should be considered

Clinical features of portal hypertension

- Faltering growth, reduced muscle bulk on overall inspection
- Splenomegaly
- Liver small, normal size or enlarged
- Cutaneous portosystemic shunts (caput medusae – flow from the umbilicus, venous hum above the umbilicus, haemorrhoids)
- Ascites

Main problems

- As a consequence of the portosystemic shunts there may be gastrointestinal haemorrhage (oesophageal varices, internal haemorrhoids) and encephalopathy
- As a consequence of the large spleen (hypersplenism) patients are often thrombocytopenic, anaemic and leukopenic

Commonest causes of portal hypertension

- Cirrhosis
- Portal vein obstruction
- Congenital hepatic fibrosis
- Hepatic vein outflow obstruction

Portal vein obstruction

In 50% there is no obvious aetiology, and it is probably a developmental defect. Other causes include congenital abnormalities of the portal vein and intra-abdominal sepsis. Presentation is usually either with asymptomatic splenomegaly or life-threatening bleeding. Occasionally patients present with faltering growth.

Congenital hepatic fibrosis

Autosomal recessive. Child looks well. Large hard liver with well-preserved liver function. Large spleen secondary to portal hypertension. Large polycystic kidneys (75%).

Differential diagnosis of portal hypertension

Having given the most likely diagnosis for your particular case, you still may be asked for some more causes of portal hypertension, so a full list is included:

Pre-hepatic

- Portal vein thrombosis

Intra-hepatic

- Pre-sinusoidal
  - Neoplasia, schistosomiasis, hepatic cyst
- Sinusoidal
  - Congenital hepatic fibrosis, cirrhosis, biliary atresia, neonatal hepatitis, alpha 1 antitrypsin deficiency
- Post-sinusoidal
  - Veno-occlusive disease

Post-hepatic

- Budd–Chiari syndrome
- Right ventricular failure
- Constrictive pericarditis

A child with portal hypertension in whom the underlying diagnosis is unclear would be referred to a liver centre for assessment. The main priority is treatment of the underlying cause.

Cirrhosis

There are numerous disorders that can lead to cirrhosis. The most likely scenario seen will be a child with biliary atresia who has had a Kasai procedure and has progressive liver disease. Other cases can be seen but rarely. The differential diagnoses should be known and will need to be presented if a child with the stigmata of chronic liver disease/cirrhosis is seen.

Tip

Remember that a cirrhotic liver is usually small (except in cystic fibrosis)

Notes on cirrhosis

Differential diagnosis of cirrhosis

Biliary tract disorders

- Biliary atresia
- Choledochal cyst
- Congenital hepatic fibrosis
- Cystic fibrosis
- Sclerosing cholangitis

Genetic and metabolic causes

- Alpha 1 antitrypsin deficiency
- Wilson's disease
- Glycogen storage disease

Infection

- Hepatitis B and C
- Cytomegalovirus

Autoimmune

Drugs and toxins

- Alcohol

Nutrition

- TPN

NB **Chronic active hepatitis** refers to inflammation, necrosis and fibrosis that can lead to cirrhosis. The commonest causes are hepatitis B and C and autoimmune liver disease.

Notes on conditions most likely to be seen

Extrahepatic biliary atresia – post-Kasai procedure

This is a very common clinical case in the examination, and patients may also be used for the history taking/management planning station. Pointers relevant to this station with regard to history taking and management planning can be found in Chapter 3, Section 3.12.

Case

What do you think of this girl's scar?

Examine the abdomen, what do you find? Can you give a sequence of events to explain this?

Clinical findings

- Characteristic scar
- Liver may be palpable
- Signs of portal hypertension may be present
- Signs of chronic liver disease may be present

Currently, biliary atresia is the commonest indication for liver transplantation in childhood. Most of this is extrahepatic biliary atresia. The incidence is 1 in 8000 to 1 in 20 000 live births. The disorder is thought to be acquired and presents as neonatal cholestasis.

Investigation is complex and any infant with prolonged jaundice that is predominantly conjugated needs to be referred to a liver centre for further advice.

Treatment is with a portoenterostomy (Kasai procedure). The outcome is much better if surgery is carried out before 3 months of age, which is why delays between finding unexplained conjugated jaundice in an infant and referring to a liver centre can be detrimental to the patient. Post-operatively there is a high risk of cholecystitis and children need intravenous antibiotic therapy if unwell. A proportion go on to get chronic liver disease and a number require transplantation.

Intra-hepatic biliary atresia

This is less common and can be either syndromic or non-syndromic. Alagille syndrome is an example worth remembering.

Alagille syndrome

- Paucity of intra-hepatic bile ducts
- Characteristic facies with growth retardation, mental retardation
- Vertebral arch defects/hemivertebrae
- Hypoglycaemia
- Heart disease – usually peripheral pulmonary stenosis

Alpha 1 antitrypsin deficiency

Autosomal-recessive phenotype determined by protease inhibitor (PI) typing. The condition presents with cholestasis in infancy, cirrhosis in childhood and chronic obstructive pulmonary disease in early adult life.

Wilson's disease

- Prevalence is 1 in 30 000. Autosomal-recessive inheritance. The pathology is as a consequence of decreased biliary excretion of copper and impaired caeruloplasmin production. Caeruloplasmin is the plasma protein that transports copper.
- Note that the condition does not present clinically until the child is over the age of 5 years, as the effects of copper build-up can take time to manifest themselves clinically.
- Younger children tend to present with liver disease – asymptomatic hepatomegaly, acute hepatitis, chronic active hepatitis, portal hypertension (ascites, oedema, variceal haemorrhage) or fulminant hepatic failure.
- An ophthalmology review should be requested as part of the work-up. Kayser–Fleischer rings are said to be pathognomonic but are not seen in every patient. Sunflower cataracts are sometimes seen.
- Other abnormalities include renal tubular disorders and haemolysis. Adults usually present with neurological symptoms secondary to copper deposition in the brain, such as a characteristic tremor and rigidity.
- Laboratory diagnosis is suggested by a low plasma caeruloplasmin level and high 24-hour urinary copper excretion. The latter can occur in other forms of hepatitis and a liver biopsy is often required. In equivocal cases the increased copper excretion after chelation with D-penicillamine is of diagnostic importance. Genetic testing is available and currently can be used for genotyping a known patient then screening first-degree relatives for that particular mutation. Untreated the condition is fatal, but treated the prognosis is good.
- Treatment is with oral penicillamine as a copper-binding agent in conjunction with a low-copper diet. Patients on penicillamine require vitamin B_6 supplements as it is an antimetabolite. Side-effects include rashes and proteinuria.
- Trientine is an alternative chelating agent with similar, but less frequent, side-effects. Zinc can also be used; it helps by reducing copper absorption from the gut.

Complications of cirrhosis

You may be asked about the main problems that a patient with cirrhosis will encounter in this station or the history taking/management planning station.

- Portal hypertension
- Bleeding diatheses
- Increased susceptibility to infections
- Ascites with danger of secondary bacterial peritonitis
- Pulmonary hypertension
- Hepatoma

- Gallstone formation
- Renal failure
- Hepatic encephalopathy
- Endocrine changes
- Impaired hepatic metabolism of drugs and hormones
- Impaired neurodevelopment

Management of cirrhosis

This is multidisciplinary and the team will be headed by a paediatric hepatologist:

- Determine the cause if possible and treat the underlying condition – it is hoped that this will minimise further liver damage
- A diet containing adequate protein and essential fatty acids, trace elements and vitamins for growth, and normal activities; plus adequate calorie intake. Specialist dietician to implement and review this regimen
- Ascites best dealt with by restricting salt intake and/or diuretics.

Emergencies – Remember A and B before C!

- Alimentary bleeding – admit, even if small, for assessment as patients may have a slow initial bleed followed by a massive bleed. Should have two reliable iv lines, and crossmatch. Further prevention and management of varices includes sclerotherapy, variceal ligation, surgical portosystemic shunts.
- Bacterial peritonitis – should be managed by adequate education of staff and parents, leading to early detection and then treatment with appropriate antibiotics.
- Hepatic encephalopathy – prevent the accumulation of ammonia, remove/correct identifiable precipitating factors, improve liver function by reducing protein intake, use antibiotics (neomycin). Lactulose to encourage excretion of waste products via the stool.
- Hepatorenal failure is usually a terminal event requiring liver transplant.

Liver transplantation

You may see a child who has had a liver transplant in this section of the exam or the history taking/management planning station. Transplant is a treatment for acute and chronic liver failure as well as for patients with metabolic disorders (particularly Wilson's disease, alpha 1 antitrypsin deficiency and galactosaemia), extrahepatic biliary atresia or liver tumour.

- Indications for assessment:
  - End-stage chronic disease – likelihood of death within 18 months or evidence of deterioration that will worsen the prognosis for liver transplantation
  - Unacceptable quality of life
  - Growth and neurodevelopmental impairment secondary to liver disease
  - Fulminant or subacute liver failure – age < 2 years or INR > 4

Transplant can also be indicated to treat recurrent, serious or life-threatening reversible complications of liver disease (eg bleeding varices, gross encephalopathy, persistent severe ascites).

- Donor:
  - A supply of suitable donors is a major limiting factor. Can split organ between recipients or use one part of the liver from a related donor (monosegment graft)
- Complications:
  - The acute rejection rate can approach 50%–60%
  - Sepsis is the primary cause of deaths post transplant as the patient needs to be pharmacologically immunosuppressed to reduce the risk of organ rejection
  - Biliary leaks occur in up to 20% of cases
- Prognosis:
  - There is around a 90% 2-year survival, and up to 80%–90% 5 year survival for planned transplants. Patients need lifelong immunosuppression. The chronic rejection rate is around 5%–10%.

10

RESPIRATORY

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10.1 Introduction

The respiratory examination is relatively straightforward and should be revised thoroughly and practised frequently. The conditions commonly seen are cystic fibrosis, other chronic suppurative lung disease, asthma and chronic lung disease (also referred to as bronchopulmonary dysplasia).

Patients with these chronic conditions are among the first to be contacted when an examination centre is recruiting families to help on exam day. Studying these conditions (approach to examination, clinical signs and knowledge of the underlying condition) in depth will present a good opportunity for you to pick up marks and boost confidence during the exam.

Some of the conditions discussed in this chapter may also come up in the history taking/management planning or the communication skills stations. The notes about these conditions will therefore be useful for other sections of the exam as well as the short cases section.

10.2 Clinical examination

Suggested approach for examination of the respiratory system

Find your own system with which you are comfortable. It is important to start with a general assessment looking for dysmorphology, nutritional status and any other specific features. The following is a suggested scheme.

Baby/infant/toddler

It is reasonable to be opportunistic in this situation and change the order of examination to gain the maximum amount of information, as long as you tell the examiner you are doing so.

Include the following:

- Observation – dysmorphology, supplementary oxygen, nutritional status, cyanosis, recession, respiratory rate, chest expansion, scars
- Auscultation (remember apices and right middle lobe)
- Apex beat and heart sounds
- Percussion (may not be helpful in infants and toddlers but should be attempted)
- Clubbing
- Anaemia
- Tracheal position
- Hepatomegaly/liver edge pushed down by hyperexpanded lungs
- Ear, nose and throat (ENT)

Toddlers may be more tolerant of the traditional order described below, but the last three steps are more likely to elicit some protest, so may be best left to the end.

Older child

- Look at the child from the end of the bed and make some general observations. Do they look underweight? Mention the need to plot their weight on a growth chart
- Children with cystic fibrosis (CF) may have a gastrostomy
- Look around the room for clues (eg inhalers/nebulisers, pancreatic enzymes, peak flow meter, sputum pot)
- Examine hands for clubbing, peripheral cyanosis (can be seen in nails) and anaemia
- Count pulse and respiratory rate – and be seen to be doing this
- Check for a BCG scar
- Assess chest shape – look for pectus excavatum/carinatum and Harrison's sulci. Might the child have kyphoscoliosis?
- While you are looking at the child's hands, arms and chest wall you may notice eczema; be prepared to comment on this as it may be relevant to your diagnosis
- Assess respiratory effort while you are still inspecting the child. Look for nasal flaring, recession, tracheal tug, abdominal respirations. Look and listen for features of upper airway obstruction (stridor, nasal congestion)
- Look in the mouth for central cyanosis
- Feel suprasternal notch for tracheal deviation
- Lift each arm in turn and look for scars (eg lateral thoracotomy) and palpate for axillary then cervical lymphadenopathy
- Assess chest expansion. Is it equal on both sides? Don't forget to look from the side for increased antero-posterior (AP) diameter as seen in obstructive airways diseases such as asthma
- Palpate apex beat (an indicator of mediastinal shift, or Kartagener syndrome if on right)
- Percuss (anteriorly, posteriorly and laterally). Practise distinguishing between resonance and hyper-resonance, dullness and stony dullness. Practise your technique for percussion; remember to explain what you are about to do and place your whole finger flush to the chest
- Auscultate (anteriorly, posteriorly and laterally). Comment on whether breath sounds are vesicular or bronchial. Be prepared to describe added sounds such as crackles (coarse or fine?) and wheezes (monophonic or polyphonic?) along with their timing
- Assess for tactile vocal fremitus, vocal resonance if appropriate (eg suspected consolidation)

At the end of your examination

- Palpate the abdomen for a liver edge (pushed down by hyperexpanded lung or genuine hepatomegaly)
- Ask to examine the ears, nose (look specifically for polyps, considering the possibility of cystic fibrosis) and throat
- You may be shown a sputum pot (whether or not you remember to ask for one!) and be asked to describe its contents
- Ask to measure peak flow rate (if appropriate)
- Ask to see a growth chart; one may be produced for comment

Different instructions may be given and you need to adapt your examination accordingly. Remember that there are 9 minutes per station so it is likely you will be asked to perform a complete systems exam. If in doubt about whether you should, talk your way through the examination:

- Examine this child's chest
- Examine this child's respiratory system
- Look at this child's chest and tell me what you see – hyperexpansion, thoracotomy scars, Harrison's sulci, wasting
- Just listen to this child's chest
- The GP says this child does not have asthma – do you agree? Perform a full respiratory examination. The examiner is expecting you to comment on distinguishing features such as Harrison's sulci and hyperexpansion, and to ask the child to perform a peak flow measurement

Notes on examination of the respiratory system

Signs of respiratory distress

Severe signs in bold are very unlikely to be seen in the exam situation but you may be asked about them:

- Audible wheeze
- Tachypnoea
- Tachycardia
- Nasal flaring
- Intercostal/subcostal/sternal recession
- Tracheal tug
- **Abdominal respirations/see-sawing of abdomen**
- **Drowsiness and confusion**
- **Cyanosis**

Clubbing

How to assess

This can be difficult if early but is often asked. We know of one candidate who was asked to stage clubbing once he recognised it. Clubbing is graded according to stages of its development:

- Stage 1: Fluctuant bogginess of the nail bed
- Stage 2: Loss of nail fold angle then occurs (Schamroth's sign); this is most easily seen by asking the child to hold their thumbs together nail to nail and looking through the gap which will have lost its normal 'diamond' shape
- Stage 3: Increased curvature of the nail bed
- Stage 4: Enlargement of the distal phalanx (later sign)

Causes of clubbing

The most common cause seen in children who have respiratory symptoms and/or signs is suppurative lung disease, but be aware of the possibility of cyanotic heart disease. It is essential to know a list of important causes for the exam.

Respiratory

- ○ Bronchiectasis/cystic fibrosis
- ○ Primary ciliary dyskinesia
- ○ Empyema/lung abscess
- ○ Pulmonary tuberculosis
- ○ Fibrosing alveolitis

Cardiac

- ○ Cyanotic congenital heart disease
- ○ Bacterial endocarditis

Gastrointestinal

- ○ Biliary cirrhosis
- ○ Chronic active hepatitis
- ○ Inflammatory bowel disease

Other

- ○ Malignancy
- ○ Idiopathic/familial

Hyperexpansion

This implies obstructive airway disease and is an extremely important physical sign. Practise the assessment of hyperexpansion. Look from the front first and then assess by looking at the child from the side and examining antero-posterior (AP) diameter and chest excursion during the respiratory phases. Comment on asymmetry if seen.

Causes of hyperexpansion – common conditions seen in the exam

- ○ Asthma
- ○ Cystic fibrosis
- ○ Bronchiectasis
- ○ Bronchopulmonary dysplasia (chronic lung disease originating in neonatal period) – look for neonatal sequelae, eg head shape, chest drain/iv scars, nasal cannulae for low-flow oxygen, gastrostomy, neurological problems
- ○ Recurrent aspiration – commoner in children with cerebral palsy
- ○ Repaired tracheo-oesophageal fistula

Tip +

Hyperexpanded chest plus clubbing implies cystic fibrosis or other chronic suppurative lung disease

Hyperexpanded chest without clubbing implies asthma or bronchopulmonary dysplasia

Harrison's sulcus

Harrison's sulcus is visible as a bilateral fixed indrawing of the anterior portion of the lower ribs. It is caused by chronic airway obstruction encouraging excessive diaphragmatic use. This eventually causes a deformity where the diaphragm inserts into the soft costal cartilage of the ribcage.

Spirometry

The routine use of lung function testing has become more widespread and most children over the age of 5 years will be able to have their lung function tested. You may be asked to interpret a test result.

Values obtained by spirometry are:

- PEFR: Peak expiratory flow rate. Effort dependent and used more for home monitoring
- FVC: Forced vital capacity. Total amount of air forcibly exhaled. Therefore FVC is reduced in restrictive lung diseases
- FEV1: Forced expiratory volume in the first second. If diminished, it indicates large- and medium-sized airway obstruction
- FEV1/FVC: Preserved in restrictive disease, lowered in obstructive diseases such as CF and asthma
- FEF25–75: Average expiratory flow between 25% and 75% of lung capacity. This provides a better indication of small airways function than FEV1/FVC. Be prepared to comment on the fact that as resistance increases, the flow-volume loop becomes more concave or 'scalloped'

It would be sensible in the run-up to the exam to attend the local respiratory clinic and discuss the results of individual patients.

10.3 Cases and conditions

Asthma

Children with asthma may be brought down from the ward to the respiratory station on exam day. This is also one of the chronic illnesses that may appear in other sections of the exam.

Instructions to candidate

Examine this child's chest.
Comment on the appearance of this child's chest.

Case

Simon is a well-looking 9-year-old boy who looks well grown for his age. I would like to plot his height and weight on a growth chart.

There is no clubbing or cyanosis. On inspection of his chest he has bilateral Harrison's sulci. His respiratory rate is 32 breaths per minute. Auscultation of his chest reveals bilateral expiratory wheeze with a prolonged expiratory component. The most likely diagnosis is asthma. I would also like to ask Simon to perform a peak flow measurement …

Clinical examination

○ Hyperexpansion, chest wall deformity (Harrison's sulci, pectus carinatum)
○ Signs of respiratory distress
○ Wheeze and prolonged expiration
○ Peak flow

Also look for: clubbing, wasting, coarse crepitations, focal signs, central lines, neonatal scars.

If these are present then an alternative diagnosis is more likely.

Tip +

Clubbing is not seen in children whose only diagnosis is asthma

Differential diagnoses of asthma – you may be asked this

- Lower respiratory tract infection
- Bronchopulmonary dysplasia (ex-prematures)
- Bronchiectasis
- Foreign body
- Congestive cardiac failure (myocarditis/congenital heart disease [CHD])
- Gastro-oesophageal reflux (severe resulting in aspiration)
- Tracheo-oesophageal fistula – post repair
- H-type tracheo-oesophageal fistula (very rare)
- Vascular ring
- Lymphadenopathy causing extrinsic small airway compression (TB/Hodgkin's/sarcoidosis)

Notes on asthma

Epidemiology

Prevalence: 10% of preschool children, rising to 30% of school-age children in some areas, depending on the definitions and questions used in prevalence studies. There has been a threefold increase in the prevalence of asthma in the past 40 years, but it is now starting to level off and even fall in some countries, including the UK.

There are still some deaths in children under 16 in the UK. The risk of death is higher if there is a history of many previous hospitalisations.

Management – summary

See Section 2.2 in History Taking and Management Planning for detailed management notes.

General measures

- The most important measure is the avoidance of cigarette smoking and also of passive exposure to cigarette smoke
- Allergen avoidance:
  - Reducing the numbers of house dust mites by washing bedding at 60 degrees, using 'anti-allergy' mattress covers, changing carpeting in bedrooms to laminate flooring
  - Avoiding animal dander

There is a theoretical benefit if the above measures are adopted, but a sustained clinical effect has not actually been proven and they are not endorsed in the latest national guideline. Nevertheless, families usually want to practise some of these measures:

- Weight reduction in obese patients is advised
- Regular exercise

Drug therapy

Keep up to date with joint British Thoracic Society (BTS) and Scottish Intercollegiate Guidelines Network (SIGN) guidelines at www.brit-thoracic.org.uk or www.sign.ac.uk. Updates are frequent.

Acute attacks

Oxygen, inhaled or nebulised beta-2 agonists, ipratropium bromide and early use of oral steroids. Nebulised magnesium sulphate can now be used in some cases. The small proportion of patients who don't respond require intravenous (iv) salbutamol or aminophylline, iv hydrocortisone and occasionally iv magnesium sulphate. A very small proportion of children need ventilating on the paediatric intensive care unit (PICU).

Long-term treatment

Summary of British Thoracic Society (BTS) guidelines for stepwise therapy

Step 1: Inhaled short-acting beta-agonist as required

Step 2: As Step 1 + regular inhaled prophylactic steroids

Step 3: Inhaled steroids plus initial add-on therapy (long-acting beta-2 agonists [LABA] +/– increase dose of inhaled steroid, or increase steroid +/– add leukotriene receptor antagonists if LABA ineffective)

Step 4: As Step 3 + increase inhaled steroid in over 5s, refer to respiratory paediatrician if under 5 (last step in this age group)

Step 5: For 5–12 year olds, as Step 4 + regular prednisolone + refer to respiratory paediatrician

Delivery devices

Remember that one of the key aspects of asthma management is educating patients and families on how to use the medication prescribed. You may be tested on this in the respiratory station by being asked to explain delivery devices to a patient, parent or examiner.

Many candidates who attend clinical courses are unable to use some of th devices correctly. This does not give the examiner a good impression. Mos hospitals have a paediatric asthma nurse who demonstrates these devices ever day and it would be wise to utilise their help before the exam! There are NIC guidelines for inhaler devices for children with chronic asthma (www.nice.org uk/TA38). The central principle of these guidelines is that the child's needs an likelihood of compliance come before cost.

Babies and toddlers – up to the age of 5 years

- **Metered dose inhaler coupled** to a **spacer device**, eg **Volumatic®** **Aerochamber plus®**. The spacer slows down the particles and requires n co-ordination to use. Even with a spacer device, airway deposition is probably still only 25%. For under 2 or 3 year-olds who are unable to breath directly into the mouthpiece, a soft rubber **mask** is supplied.
- Spacers must be replaced when they become scratched. When they ar cleaned they must be left to drip-dry, not wiped with a cloth, as the stati charge produced reduces effective particle delivery to the lungs. While mos toddlers cope well with these devices, babies tolerate them less well.

Age 5–15 years

- Pressurised metered dose inhaler **and** spacer device for corticosteroi therapy.
- **Self-activating devices with propellant** such as **Easibreathe®** an **Autohaler®** devices. The child still needs to be able to take a deep breat and hold it when using them. Inhalers should now be chlorofluorocarbo free.
- **Dry powder inhalers** such as the **Accuhaler®** and **Turbohaler®** are simpl to use and are popular with school children. **Diskhalers®** can be very usef and are easy to carry to school. You will need to learn the technique c loading and using this inhaler as it is different from all the others.

Bronchiolitis

Case

> *Jade is 6 months old. She appears to be well grown. I would like to plot her on a growth chart. She is being treated with oxygen via nasal cannulae and is in respiratory distress with tachypnoea of 60 breaths per minute, nasal flaring and intercostal recession. There is no cyanosis. Auscultation of her chest reveals bilateral inspiratory and expiratory wheeze and bilateral fine crepitations. The most likely diagnosis is bronchiolitis ...*

Clinical examination

- ○ Hyperexpanded, barrel-shaped chest, nasogastric (NG) tube, low-flow oxygen
- ○ Signs of respiratory distress – mainly tachypnoea, recession
- ○ Cough – often has a characteristic dry, wheezy quality
- ○ Nasal discharge
- ○ Fine inspiratory crackles
- ○ Expiratory wheeze
- ○ Air entry may be unequal due to consolidation caused by atelectasis (around 30% of patients)

Differential diagnoses include

- ○ Heart disease: ventricular septal defect/atrioventricular septal defect, myocarditis, total anomalous pulmonary venous drainage
- ○ Cystic fibrosis
- ○ Bronchopneumonia, particularly staphylococcal or pneumococcal. Patients with a high fever (axillary temperature of > 39°C) should be assessed for features of bacterial infection
- ○ Aspiration
- ○ Pertussis
- ○ Post-viral wheeze
- ○ Foreign body

Notes on bronchiolitis

Bronchiolitis is a seasonal viral respiratory illness that most commonly presents in infants between 3 and 6 months of age. It occurs in winter epidemics in both hemispheres. The main causative agent is respiratory syncytial virus (RSV) in around 75% of cases (serotypes A and B). Some other common viral causes are rhinovirus, influenza, parainfluenza, adenovirus and human metapneumovirus. The underlying pathophysiology is inflammation of the small airways (bronchioles). Infants are at increased risk of hospitalisation if their parents smoke and if they live in overcrowded conditions. Breastfeeding (especially if continued for over 2 months) is partially protective, presumably through passive transfer of anti-RSV immunoglobulin.

The illness starts with a few days of coryza followed by a moist cough and sometimes wheeze. The child may have a fever and many go through a phase of feeding poorly. Very young or ex-premature infants can present with or develop apnoeas. The diagnosis is clinical, but the guidelines published by the Scottish Intercollegiate Guidelines Network (SIGN) recommend rapid testing for RSV (by immunofluorescence of a nasopharyngeal aspirate) to aid decisions about cohorting for those patients who require admission.

Management

For more information the SIGN guidelines on bronchiolitis are available from www.sign.ac.uk, guideline no. 91. There are new NICE guidelines being developed at the time of writing.

Mainstay is **supportive care with frequent monitoring**:

- Supplementary humidified oxygen by headbox or nasal cannulae; mandatory if saturations in air in the acute phase of the illness are less than 92%. Additional respiratory support may be required either by continuous positive airway pressure or transfer to a PICU if ventilation is required. Bronchiolitic patients can be challenging to ventilate.
- Hydration and providing help with feeding. Nasogastric feeds (small, frequent amounts) are more physiological but may not be tolerated very well. Intravenous fluids may be required, with monitoring of electrolytes, bearing in mind that many patients develop the syndrome of inappropriate antidiuretic hormone (SIADH).
- Suction (gentle) and saline nose drops can be useful as secretions are usually profuse and contribute to the airway obstruction and poor feeding.

Pharmacotherapy

- You may be asked about the role of pharmacotherapy. **The vast majority of patients do not benefit from any drugs and this should be stated to the examiners if you are asked.**
- It is useful to be aware of the various therapies that are being considered and trialled as you may need to know this information in the history taking and management planning or communication skills stations. It is prudent to look at the latest incarnation of the SIGN and NICE guideline when considering which therapies are recommended at the present time.
- Hypertonic (3%) nebulised saline has been proposed, although use is controversial and there is only limited evidence. It should in theory reduce mucous plugging and settle airway oedema.
- **Ipratropium bromide** via nebuliser reverses some of the short-term effects of bronchiolitis but has not been shown to alter mortality or length of hospitalisation.
- There has been considerable interest in the use of **nebulised adrenaline** but it is currently not recommended.
- **Montelukast** has been tried, although there is insufficient evidence to recommend its routine use.
- **Nebulised ribavirin** was found to reduce viral shedding and shorten length of hospital stay in one study. However, systematic reviews have shown no reduction in mortality. It is not recommended unless the patient is in a very high-risk group.
- **Steroids** are of no benefit during the acute phase. They are of some use in obliterative bronchiolitis. This is a destructive form of bronchiolitis, usually

associated with adenovirus, which becomes chronic and results in bronchiectasis.

- ○ **Antibiotics** are of no benefit in straightforward bronchiolitis but are occasionally required if there is clinical suspicion of super-added bacterial infection – around 5%–10% of patients in some centres.
- ○ **Palivizumab**, an anti-RSV humanised monoclonal antibody, is available for **prophylaxis** against severe illness in high-risk patients (see below). However, it is very expensive, so cost-effectiveness is an issue. It is only recommended for patients within these high-risk groups.

High-risk groups

The threshold for admission should be lowered in the following groups:

- ○ Congenital heart disease
- ○ Cystic fibrosis
- ○ Chronic lung disease (ex-premature babies at higher risk even if they do not have CLD)
- ○ Immunodeficiency

Discharge

Consider when a baby is obviously recovering clinically, and has sats of > 92%–93% in room air for over 8–12 h, including during a period of sleep, and is taking over 75% of feeds.

The social set-up is also relevant. On discharge parents should be warned that some infants take up to 4 weeks to recover as they are left with inflamed, hyper-reactive airways that contain damaged cilia. Such infants can re-present multiple times to their GP or paediatric unit with episodes of wheeze. These patients very rarely benefit from steroid prophylaxis. There are some infants who take even longer than this to recover, particularly if they are passively exposed to smoke.

Cystic fibrosis

Children with cystic fibrosis are seen very commonly in the respiratory station. A thorough knowledge of all aspects of this condition is required for this *and* for other parts of the exam. Remember, cystic fibrosis is a multisystem disorder.

Instructions to candidate

Examine this child's chest – central line in situ, hyperexpansion, crackles/wheeze.

Examine this child's hands – clubbing.

Look at this child – wasting/nasogastric tube, height and weight.

Examine this child's chest – what else would you like to examine? (Abdomen, nutritional status, ENT, cardiac).

General examination

Key observations

- ○ Around the bed – pancreatic enzymes, peak flow meter and nebulisers in evidence
- ○ Visible central line or gastrostomy
- ○ Cyanosis, supplemental oxygen
- ○ Wasting – reduced muscle bulk (mid-arm circumference), reduced subcutaneous fat (skinfold thickness)
- ○ Obvious hyperexpansion and respiratory distress
- ○ Scars – thoracotomy, abdominal

Remember to measure or ask about height and weight, and offer to plot these on a centile chart.

Case

John is a 15-year-old boy who is lying in bed, comfortable at rest. There is a central line and gastrostomy tube in situ. He looks thin with a poor muscle bulk and reduced subcutaneous fat. I would like to plot him on a height and weight chart. Looking at his hands he appears clubbed.

He is obviously hyperexpanded and using his accessory muscles of respiration. His respiratory rate is 24 breaths per minute …

On examination

Hands/face

- ○ Clubbing – in older child, CF unlikely in the absence of clubbing
- ○ Peripheral and central cyanosis
- ○ Pulse
- ○ Pallor
- ○ Jaundice
- ○ Nasal polyps

Chest

- ○ Tachypnoea
- ○ Hyperexpansion
- ○ Crackles, wheeze
- ○ Peak flow
- ○ Sputum sample (ask at end)

Abdomen

- ○ Scars – meconium ileus
- ○ Gastrostomy
- ○ Caput medusae
- ○ Ascites
- ○ Hepatosplenomegaly
- ○ Rectal prolapse
- ○ Palpable faecal mass – distal intestinal obstruction syndrome (DIOS) or meconium ileus equivalent

Cardiovascular system

- ○ Signs of right-sided heart failure secondary to pulmonary hypertension (raised JVP, dependent oedema, hepatomegaly)

Endocrine system

- ○ Poorly grown, delayed puberty
- ○ Insulin injection sites

Joints

- ○ Arthritis – occurs in 1% of patients and is fleeting. Knees and ankles most commonly affected. Thought to be due to circulating immune complexes.

> ***Tip*** +
>
> Patients with multisystem involvement may be asked to cover other stations, and so you may get asked to examine systems other than respiratory in the child with cystic fibrosis

Notes on cystic fibrosis

Inheritance is autosomal recessive. Prevalence 1:2500, carrier frequency 1:25. Cystic fibrosis transmembrane conductance regulator (CFTR) is a cAMP-mediated chloride channel. In the lungs, normal CFTR causes efflux of chloride ions across apical membranes of the submucosal glands. Sodium and water follow. Abnormal CFTR (reduced production, defective processing or ATP-binding site defects) causes an inability to secrete chloride, sodium and then water, resulting in viscid secretions, the hallmark of CF. A similar problem occurs in the pancreatic and biliary ducts, and also in the reproductive tract. The inspissated mucus in the bronchi causes failure to clear pathogenic organisms. These then colonise the airways and the resulting inflammatory reaction predisposes to repeated infections and chronic suppurative lung disease. However, in the skin the function of the sweat glands is to absorb chloride from the isotonic sweat. CFTR defects therefore cause a high sweat chloride, the basis of the sweat test.

Genetics

There are now almost 2000 alterations at the gene locus on chromosome 7. Delta F508 is the commonest mutation and is present in 75% of patients in the UK. Two-thirds of cases are homozygous, one-third heterozygous for Delta F508 and one other mutation. Different phenotypes exist, and the genotype/phenotype correlation is strongest for pancreatic insufficiency and sweat chloride. For example, all Delta F508 patients have pancreatic insufficiency whereas those with the R117H mutation have some preservation of pancreatic function.

Diagnosis

- Antenatal chorionic villous sampling (at 8/40 to 10/40), amniocentesis (at 16/40 to 18/40) for gene analysis if mutation within the family has been previously identified.
- Guthrie test looking at immunoreactive trypsin (IRT), which is a reflection of pancreatic damage. A single raised IRT (taken as > 99.5th centile) has low specificity when used alone, so genetic testing is then performed on the sample. If two genes are found, repeat genetic testing is advised. If one gene is found, the child is referred for a second blood spot IRT. A subgroup of patients who have a high false-negative rate are those with meconium ileus. Genetic analysis is strongly recommended irrespective of the IRT result. Screening and so early diagnosis have been shown to help to optimise growth and nutritional status – an important aspect of management. The UK National Screening Committee recommends that all babies in the UK should be offered screening for cystic fibrosis. Remember that screening may miss a small percentage of cases. Clinical presentation is very important and may lead to more in-depth testing for rarer mutations.

- Genetic analysis – in all children who have been diagnosed, as this provides more information on genotype/phenotype correlations. Also used when sweat test is equivocal.
- Sweat test using pilocarpine iontophoresis. Sweat is collected over a period of about 30 minutes. A sweat chloride concentration of > 60 mmol/l supports the diagnosis. Over 100 mg of sweat are required for the filter paper technique, but newer methods are being developed that require smaller amounts. If the test is abnormal (and no genetic tests have been performed) it should be repeated.

Clinical problems in cystic fibrosis – summary

More detailed notes on the following problems and their management can be found in Chapter 2.

Respiratory

- Bacterial colonisation and infection – typically *Staphylococcus, Haemophilus, Pseudomonas*
- Pulmonary hypertension
- Allergic bronchopulmonary aspergillosis (ABPA)

Gastrointestinal/hepatic

- Pancreatic insufficiency with resultant malabsorption, especially of fat (85%)
- Meconium ileus in neonates (10%–15%)
- Distal intestinal obstruction syndrome (DIOS) or meconium ileus equivalent in older patients
- Rectal prolapse (toddler presentation)
- Chronic liver disease – mainly in teenagers (around 20%) and usually resulting from biliary cirrhosis

Endocrine

- Insulin-dependent diabetes in 7% of paediatric patients (usually over 10 years of age). The incidence increases in adulthood, with around 50% of patients having CF-related diabetes by the time they are 30 years of age.

Metabolic

- Salt deficiency state: hyponatraemic, hypochloraemic metabolic alkalosis

Genitourinary

- In boys, infertility caused by azoospermia due to bilateral absence of vas deferens. Some women are subfertile

Management of cystic fibrosis

Multidisciplinary and often outpatient-based. Annual review advised:

- **Physiotherapy** (a minimum of twice daily) to minimise the impact of airway secretions
- **Nutrition** must be optimised because patients with cystic fibrosis have increased metabolic demands, fat malabsorption and poor appetite. A paediatric dietitian should be involved. A high-calorie, high-protein diet (around 120%–150% of that for an unaffected individual) is required, with regular plotting of weight for height and BMI. Some patients require short periods of nasogastric feeding, but if consistent weight gain is difficult to achieve, gastrostomy feeding is used
- **Pancreatic enzyme supplements** (pancreatin) in pH-sensitive polymer capsules or, for infants, granules that can be mixed with feeds
- **Vitamin A, D and E supplementation** is necessary, as these are fat soluble. Plasma concentrations should be measured at annual review, and prothrombin time should also be checked as a marker of whether vitamin K supplementation is required as well
- **Chest infections** – be aware that 3-monthly surveillance using cough swabs or preferably sputum cultures is recommended, regardless of current health. Be vigilant about checking for *Burkholderia cepacia*:
    - Flucloxacillin is used either prophylactically from diagnosis (mainly in first 2 years of life) or long-term after colonisation with *Staphylococcus*
    - **Nebulised tobramycin/colomycin** used in *Pseudomonas* colonisation
    - Acute infections are treated aggressively as they arise, often using the results obtained from the last cough swab/sputum samples
    - Oral (eg **ciprofloxacin**) or intravenous (eg **ceftazidime/piperacillin** plus an aminoglycoside such as **tobramycin**) therapy can be started as appropriate
- Nebulised hypertonic saline: reduces the viscosity of secretions
- **DNase**: To digest viscous DNA fragments derived from inflammatory cells. This is a truly evidence-based treatment, the high expense of which is justified so long as other therapies are optimal
- **Treatment of co-existent conditions/complications:**
    - Asthma: bronchodilators
    - ABPA: corticosteroids
    - Constipation and DIOS: laxatives and hydration, through to Gastrografin® or even surgical treatment
    - Arthritis: NSAIDS, prednisolone

  - Diabetes: insulin
  - Liver disease: ursodeoxycholic acid
  - Cor pulmonale: heart–lung transplant
- Infection control measures, including separation of patients in clinic settings: to reduce spread of harmful organisms such as *Burkholderia cepacia*
- Psychological and social support
- Gene therapy: be aware of this although still in early stages.

Bronchiectasis

This usually follows airway obstruction that has been sufficiently prolonged to cause infection of the retained secretions. The net result is a dilated, chronically inflamed airway. The lower lobe is most commonly affected, followed by the middle and then the upper lobe. The chest presentation is as for cystic fibrosis, and the latter is part of the differential diagnosis. Other causes are discussed here. Bronchiectasis is often first noted on chest X-ray and confirmed using high-resolution CT with or without bronchoscopy.

Aetiology

Most cases are post-infectious, due to immune deficiency, or are idiopathic; these should be mentioned first:

- Post-infectious: mainly staphylococcal and streptococcal pneumonias, adenovirus, pertussis, influenza
- Primary ciliary dyskinesia
- Bronchiolitis obliterans (adenovirus, RSV, pertussis)
- Alpha 1 antitrypsin deficiency
- Asthma
- Bronchial compression caused by airway lymphadenopathy or, more rarely, by a tumour
- Recurrent aspiration (H-type tracheo-oesophageal fistula or TOF)
- Lobar sequestration:
  - A non-functioning mass of embryonic tissue within the chest that receives its entire arterial supply from the aorta
  - Haemoptysis is common
  - There is a continuous murmur over the lesion
  - Needs aortography before removal because feeding vessel usually arises from abdomen
- Immunodeficiency:
  - Hypogammaglobulinaemia
  - IgA deficiency
  - Complement deficiency
  - Hyper IgE (Job) syndrome
  - AIDS

- Syndromic:
  - Yellow-nail syndrome (grossly abnormal nails, lymphoedema)

Management

In early cases the condition can be reversed, so it is important to manage it aggressively.

- Education; aids compliance with treatment
- Regular physiotherapy for life; encourage exercise where appropriate
- Prophylactic antibiotics for most patients
- Bronchodilators as bronchospasm is usually a prominent feature
- Mucolytics when appropriate, although there is no clear benefit from DNase
- Prompt immunisation – including influenza

Monitoring

Regular clinic appointments to look at following:

- Spirometry (often normal, with an emerging obstructive picture as the condition becomes more severe)
- Sputum cultures to detect colonising organism and its sensitivities
- Monitor for cor pulmonale
- Growth and nutrition are very important (can suffer due to a combination of increased metabolic rate and reduced intake). Offer nutritional supplements if necessary or feed in alternative ways, eg overnight nasogastric or gastrostomy feeds

Surgical treatment

- Lobectomy may be necessary – involve paediatric surgeons early as need a window to operate when the child is clinically well enough for the procedure

Primary ciliary dyskinesia (PCD)

This is a mainly autosomal-recessive group of disorders in which cilial movement is inefficient or absent.

Case

> *Peter is a well-looking 7-year-old boy who appears small for his age and I would like to plot him on the appropriate height and weight centile charts. On examination he has clubbing, and is not cyanosed. Palpation of his apex beat reveals dextrocardia and auscultation of his chest reveals coarse crepitations at both bases. The most likely diagnosis is Kartagener syndrome. I would like to examine the abdomen, enquire about a history of sinusitis …*

Features of primary ciliary dyskinesia

- Bronchiectasis
- Nasal polyposis
- Chronic sinusitis
- Chronic suppurative otitis media (may have a hearing aid, preferred to grommets in this condition)
- Male infertility (common)

Up to half of all patients with PCD have Kartagener syndrome with all of the above features plus:

- Dextrocardia (palpating for the apex beat, which will not be on the left *but on the right*, should alert you to this diagnosis!)
- Visceral situs inversus

Notes on ciliary dyskinesia

There is a defect in cilial ultrastructure, mobility or both. The most obvious effect is on the mucociliary escalator, which depends on cilia wafting mucus from the lower airways in a cephalic direction from where it is usually swallowed. The cilia look grossly normal but have ultrastructural defects. Primary ciliary dyskinesia occurs in 1:15 000 of the population, accounting for as much as 5% of children with recurrent respiratory disease. It usually presents with bronchiectasis after a troublesome period of chronic wet cough, sinusitis, nasal polyps and chronic suppurative otitis media. This condition should be considered in neonates with unexplained persistent tachypnoea, pneumonia with no risk factors and of course, dextrocardia with situs inversus. There are gastrointestinal associations such as trachea-oesophageal fistula, oesophageal atresia, biliary atresia and volvulus. Hydrocephalus is commoner; there are cilia lining the ventricles that can be compromised by PCD.

Diagnosis is made and management implemented with the help of specialist diagnostic services based in a small number of UK centres.

An initial screening test which is sometimes attempted involves placing a drop of saccharin on the nasal mucosa and measuring the time that elapses before the patient reports the sensation of a sweet taste in the mouth (normally 11 minutes, and it should be less than 1 hour). The patient must sit still for an hour and not sniff, cough, eat or drink, which precludes the testing of many younger children!

Patients as young as about 4 years can be screened using nasally exhaled nitric oxide measurements, which are low in PCD.

Assessment of ciliary structure and function is the best diagnostic test. Nasociliary brushings from turbinates are obtained, and motility is assessed in vitro by photometry, giving a beat frequency and a comment on whether movement is dyskinetic. Electron microscopy will reveal ultrastructural abnormalities.

Prognosis: some patients have a normal lifespan, especially if they are diagnosed relatively early so that lung disease progression is modified (by educating patient about avoiding smoking/allergens and teaching them physiotherapy) and recurrent infections treated. Annual influenza vaccinations are recommended, on top of the normal immunisation schedule. Hearing should be regularly monitored; some children can have significant hearing loss requiring hearing aids.

Chronic lung disease of infancy

This is a very common short case and a typical 'crossover' case with the history taking and management planning station (see Section 2.3). Another term for this particular condition is bronchopulmonary dysplasia (BPD).

Case

Jordan is 6 months old but looks small for his age. I would like to plot his weight, length and head circumference on a centile chart. He has nasal cannulae in situ which are connected to an oxygen cylinder. He also has a long, narrow head shape.

There is no clubbing. He has a hyperexpanded chest. There is mild intercostal recession. He has two chest drain scars on the right and the dorsum of both hands have venepuncture scars. Auscultation of the chest reveals bilateral wheeze. The most likely diagnosis is chronic lung disease.

I would like to perform a developmental examination …

Notes on chronic lung disease

The definition of chronic lung disease states that the child must have required respiratory support with or without ventilation after birth and still be oxygen dependent at 28 days with evidence of respiratory distress and an abnormal chest X-ray. The incidence is rising as survival of extremely premature babies improves. Prevalence is around 20% of all ventilated babies.

Associations: high levels of inspired oxygen, barotrauma and volutrauma, patent ductus arteriosus (PDA), air leak.

Clinical features

- General appearance will be of an infant who is obviously small for chronological age
- Head may have a characteristic scaphocephalic appearance and the face may look cushingoid (dexamethasone). Scaphocephaly refers to a long and narrow head shape (literally means 'boat-shaped') and can occur from postnatal moulding in preterm infants or more rarely from premature closure of the sagittal suture
- Chest drain or venesection scars should be looked for, and a thoracotomy scar suggests previous PDA ligation
- Chest is usually hyperexpanded
- Comment on the presence of cyanosis (unlikely in the exam), or respiratory distress
- Stridor (secondary to prolonged intubation) or wheeze (reduced small airway diameter) may be present
- Ask to examine the heart, in particular, for a PDA or a loud second heart sound of pulmonary hypertension
- There may be an abdominal scar for a Nissen's fundoplication or gastrostomy
- Last of all, mention that you would go on to perform a developmental examination

Differential diagnoses of this picture

- Other causes of hyperexpanded chest/bronchiectasis
- Immunodeficiency, eg severe combined immunodeficiency/chronic granulomatous disease
- Chronic aspiration due to gastro-oesophageal reflux/repaired tracheo-osophageal fistula
- Congenital heart disease (eg atrioventricular septal defect), cardiac failure

Treatment

Current best evidence suggests the following:

- Supplemental oxygen while avoiding hyperoxia in extremely preterm infants; if ventilated, try and avoid volutrauma and barotrauma. Volume-targeted ventilation can help achieve this.
- Nutrition – additional calories are essential to combat increased energy expenditure and maximise alveolar growth.
- Dexamethasone if necessary – proven benefit in weaning off ventilator or decreasing oxygen requirement.

  However, while glucocorticoids can improve lung function, there are significant potential complications including hyperglycaemia, hypertension and intestinal perforation. They have not generally been shown to improve mortality. Some long-term follow-up studies have raised concern about an increased rate of cerebral palsy in patients given glucocorticoids, although confounding factors make this difficult to interpret. Current practice is to reserve glucocorticoid treatment for those who are on maximum ventilatory support with a high oxygen requirement and not showing signs of improvement, with no other causes found for this predicament. Because of the possible side-effects on neurodevelopmental outcome, this decision is always discussed with the parents and the wider clinical team. If glucocorticoid treatment is started, remember to mention the need to check blood pressure and blood glucose measurements daily.

- Diuretics – also shown to have short-term benefits in improving lung function, but there is no evidence that they affect long-term outcome. Increase risks of nephrocalcinosis and electrolyte imbalance. Generally not recommended.
- Gastro-oesophageal reflux is frequently present and a trial of therapy is worthwhile in difficult cases.
- Assess and treat complications (eg PDA, right-sided heart failure, osteopenia of prematurity).

Prognosis

- Most patients are weaned off oxygen before discharge. If discharged home with oxygen, most are weaned off by 1 year of age
- Long-term respiratory function tends to recover, but many patients are prone to cough and wheeze in infancy (tends to respond poorly to bronchodilators) and early childhood
- High risk of hospital admission with intercurrent infections
- Remember vaccinations including influenza and palivizumab (passive immunisation to protect against RSV)
- Nutrition and growth need to be carefully monitored; some babies need more calorie-dense feeds
- Neurodevelopmental follow-up assessment is recommended

Kyphoscoliosis

Children with this problem may be under a respiratory team, but could be asked to attend for the musculoskeletal/other, neurological or the history taking and management planning station.

As well as performing a thorough respiratory examination, inspect from behind. Check shoulders for asymmetry of rotation and comment on whether they are level. Palpate the course of the spine. If the child's condition permits them, ask them to bend forward. Scoliosis is positional if disappears on bending down, otherwise it is structural and pathological.

Aetiology

Commonly

○ Idiopathic – usually more severe in prepubertal girls. Often termed adolescent idiopathic scoliosis (AIS)
○ Cerebral palsy

Others

○ Duchenne muscular dystrophy
○ Spinal muscular atrophy (type II/III)
○ Friedreich's ataxia
○ Poliomyelitis
○ Neurofibromatosis type I
○ Homocystinuria
○ Osteogenesis imperfecta
○ Marfan syndrome
○ Klippel–Feil sequence (fused cervical vertebrae, webbed neck, facial asymmetry, deafness, ventricular septal defect, renal anomalies, Sprengel shoulder)
○ Alagille syndrome (broad forehead, prominent chin, posterior embryotoxon, peripheral pulmonary artery stenosis, butterfly vertebrae, cholestasis)
○ Mucopolysaccharidoses (short, large head, macroglossia, hepatosplenomegaly, cloudy cornea in Hurler's)

Notes on kyphoscoliosis

Scoliosis is lateral (coronal) curvature of the spine accompanied by rotation of the vertebral bodies and viscera. Rotation to the right is commoner than to the left.

Kyphosis is excessive antero-posterior (sagittal) curvature. Both can be present in the same patient, to varying degrees. Back pain and cosmetic issues are often of most concern to patients.

Very severe thoracic scoliosis causes a reduction in chest wall compliance and its major importance is reduction of lung volumes causing a restrictive defect. The earliest manifestation of this is a fall in PaO_2 due to ventilation–perfusion mismatch at the bases. Later, hypoventilation results in hypercapnia.

Finally, cor pulmonale can develop in response to pulmonary hypertension.

Investigation includes plain X-rays to demonstrate the angle of the scoliosis (called the Cobb angle, which correlates well with the severity of reduction in lung volume) and lung function testing to assess the restrictive defect.

Overnight oxygen saturation monitoring can be used to assess the degree of hypoxia, and is often combined with carbon dioxide studies in severe cases.

Treatment involves monitoring for complications and treating as appropriate. Bracing is suitable for some children who are still growing. There is the eventual need for corrective orthopaedic surgery in some. Surgery is a major undertaking that always requires careful consideration, as there is the potential for many post-operative complications. The benefits are mainly cosmetic, with a reduction in back pain also reported by most patients.

Prognosis depends on the cause. Idiopathic scoliosis usually has a good outcome; outcome of scoliosis due to other causes depends on the prognosis of the underlying condition.

Stridor

It is just possible that you might see a (stable) child with stridor, which is a 'crowing' inspiratory noise due to upper airway obstruction. Stridor is usually inspiratory, which suggests extra-thoracic obstruction, but can be biphasic, which suggests intra-thoracic obstruction.

Instructions to candidate

Look at this child's chest and tell me what you see and hear; what is the most likely reason for this?

What investigations are appropriate?

Case

John is a well-looking 2-month-old infant who looks well grown for his age, although I would like to plot him on a growth chart. There is audible stridor with slight sternal recession. There is no tachypnoea. There are no scars and I cannot see any haemangiomas on inspection of his skin. Chest expansion looks normal and there is no abnormality on auscultation. (Ask mother about feeding.) He is currently feeding without any problems. The most likely diagnosis is laryngomalacia, but I would also consider the following in my differential diagnosis ...

Notes on stridor

The commonest cause of stridor is laryngomalacia or 'floppy larynx'. This condition presents soon after birth. If present at birth an alternative diagnosis should be sought. It is characteristically better when the child is laid prone. It normally has a benign course and resolves by 18 months. Some neonates and infants run into problems with feeding, and a combination of poor intake and increased energy expenditure (accessory muscle use) may necessitate nasogastric tube feeding for a while. Patients sometimes need hospital admission during intercurrent respiratory infections.

Other causes to exclude:

- In a sick pyrexial child: acute epiglottitis, bacterial tracheitis, severe croup
- Inhaled foreign body
- Anaphylaxis
- Hypocalcaemia
- Hereditary angioedema (C1 esterase inhibitor deficiency)

Chronic causes other than laryngomalacia:

- Prolonged neonatal intubation – subglottic stenosis
- Tracheo/bronchomalacia
- Pierre Robin sequence
- Intraluminal web or haemangioma
- Extrinsic compression from vascular ring (double aortic arch, pulmonary sling or aberrant subclavian artery), lymphoma, branchial/thyroglossal cyst, cystic hygroma

Investigations

In children with laryngomalacia in whom a confident diagnosis can be made and there are no complications (eg faltering growth, apnoea, respiratory distress, hoarseness), no investigations are required.

In children in whom there is doubt about the diagnosis, or a biphasic stridor or an underlying cause is suspected, ensure that calcium levels are normal. A chest X-ray, looking at the mediastinum and vascular structures, should be taken. Further investigations should be discussed with paediatric respiratory, ENT and radiology teams but might include:

- Barium swallow (extrinsic compression may be seen)
- Transoesophageal ECHO (suspected vascular ring)
- Thoracic MRI (if extrinsic compression on chest X-ray/barium)
- Fibre-optic laryngoscopy and bronchoscopy

Older children with chronic stridor should also undergo lung function testing.

Miscellaneous conditions

Pierre Robin sequence

This sequence is caused by posterior attachment of the tongue (genioglossus) together with a small mandible. This results in glossoptosis and pseudomacroglossia. The tongue commonly falls back and obstructs the airway. There is sometimes a cleft palate. The mandible usually grows sufficiently within 6 months, but some children with severe recurrent obstruction will need either a nasopharyngeal airway or tracheostomy while this occurs.

Enlarged tongue

May also be seen in the 'other' station. Causes:

- ○ Trisomy 21
- ○ Hypothyroidism (including congenital)
- ○ Mucopolysaccharidoses/GMI gangliosidosis
- ○ Beckwith–Wiedemann syndrome
- ○ Pseudomacroglossia

Tracheostomy

Patients with a tracheostomy are usually well known to the examination centre and those who are well enough to be at home are often used for a few sessions on exam day. Their background condition may mean they can crop up in other stations. Management of blocked tracheostomy may be asked – please refer to the latest edition of *Advanced Paediatric Life Support – The Practical Approach* (An ALSG manual published by the BMJ group).

Indications

- ○ Obstructing mass such as cystic hygroma or haemangioma
- ○ Subglottic stenosis (high tracheostomy)
- ○ Laryngomalacia (severe)
- ○ Tracheomalacia or bronchomalacia
- ○ Prolonged intubation
- ○ Irradiation or burns
- ○ Neuromuscular weakness
- ○ Severe sleep apnoea (rare)
- ○ Severe gastro-oesophageal reflux (rare)
- ○ Long-term respiratory support (many potential indications)

Complications

- ○ Hypoxia – due to partial (secretions) or complete obstruction
- ○ Decannulation
- ○ Ulceration
- ○ Infection
- ○ Bleeding
- ○ Arrhythmias (during suctioning)

11

NEUROLOGICAL/ NEURODISABILITY

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11.1 Introduction

Paediatric neurology is a challenging and varied subject. There is potentially a huge amount of written information about the neurological examination itself, let alone the various conditions that could be seen. We have largely confined ourselves to providing a practical outline of how to examine patients in different scenarios, and describing the commoner signs and conditions in sufficient detail for the candidate to remember important points.

We would recommend further reading about the neurological examination, as the information presented here is intended to be a summary, and neurological signs can change in relation to the age of the child. This will help you with your preparation for the examination. The most important thing to do is to **practice often, so that performing the examination becomes automatic,** and you can then concentrate on how you would present the abnormal findings that you discover.

11.2 Cranial nerves and eyes

The cranial nerves and the eyes have been included together because of the considerable overlap between the two subjects; they are best revised with this in mind.

Tip +

Patients with eye signs may also be in the "other" clinical station

Cranial nerve examination needs to be confident and it is essential to develop an examination scheme and practice it with colleagues.

It is essential also to practise so you are familiar with examination of the eyes. It is particularly important to remember that early on in the examination some assessment of visual acuity needs to be made. You may be asked to examine a child who is blind; do not be caught out by this. The best way to practise eye examination and to learn about the common eye conditions is to sit in on clinics with the orthoptists and ophthalmologists. This will ensure that you are clear about the formal assessment of visual acuity and can quickly and competently assess a squint.

Suggested scheme for examination of the cranial nerves – older child

This is a very commonly examined topic and your technique must be perfect. It is essential to practise cranial nerve examination on patients and, if necessary, on colleagues. In the past you were unlikely to be asked to examine all the cranial nerves in one case but now there are 9 minutes to fill. Be prepared for many different scenarios!

Tip

Always inform the child what you are about to do and if possible, why

- Enquire about sense of smell (**olfactory nerve**), and be prepared to examine formally if requested
- Test visual acuity (**optic nerve**). Establish if the child can see (both eyes). If the child wears glasses offer to test the visual acuity with them on. This can be done grossly at the bedside by asking the child to read from an age-appropriate book, or to read a poster on the wall
- Comment on any obvious abnormality such as squint and go on to examine formally if present
- Examine visual fields – test confrontation:
    - Sit with your face opposite the child's, 1 metre apart. Cover your left eye and ask child or parent to cover child's right eye. Move your finger in from the periphery at around 2, 4, 8, 10 o'clock and 'wiggle' it. Ask the child to say 'yes' when they can see your finger 'wiggling'. Test both eyes.
- **Examine eye movements (cranial nerves III, IV, VI)** – this is really very simple as long as you remember the patient's perspective. Ask the child to follow your finger/toy with their eyes. Keep the object being followed approximately 0.5 m from their face. The plane of movement should be parallel to the child's face. Keep your other hand on the child's forehead to steady it. Ask the child to tell you if they can see two of the object. Test lateral gaze and then vertical gaze

Tip

A common pitfall is to test to the extremes of vision, where most of us will have diplopia

- Check for nystagmus – present at extremes of gaze
- Test the **sensory divisions of the trigeminal nerve**. Ask the child to shut their eyes and to say 'yes' when they feel your touch. Gently touch the face at irregular intervals with a wisp of cotton wool, on each side, in each division of the nerve – ophthalmic, maxillary and mandibular. Ask the child if it feels the same on both sides

- Test the **motor component of the trigeminal nerve (V)** by asking the child to close their jaw tightly and palpate temporalis and masseters. Mention the jaw jerk reflex and perform this in older children
- Mention corneal reflexes at the end; you are unlikely to be asked to test these
- Test the **motor division of the facial nerve (VII)**. Ask the patient to raise their eyebrows, puff out their cheeks and smile. Look for asymmetry. Remember the sensory division, which supplies the anterior two-thirds of the tongue with taste – not usually necessary to test
- Test the **auricular nerve (VIII)** grossly by asking the child if they can hear a very quiet noise that you make in each ear, eg whisper a number the child can repeat and occlude the other ear. Offer and know how to test formally (see Chapter 13). Ask parent about hearing
- The **glossopharyngeal nerve (IX)** can be tested by the gag reflex. This should not be attempted in the exam but do mention it. Remember that the gag reflex tests the glossopharyngeal (sensory) and vagal (motor) nerves. The glossopharyngeal nerve also supplies the posterior one-third of the tongue with sensation
- Test the **vagal nerve (X)** by asking the child to say a short sentence. The voice will be hoarse if there is a vocal cord paralysis. The vagus can also be assessed at this point by asking the child to say 'ah' and looking at the soft palate
- Test **accessory nerve (XI)** by forced rotation of the head to the contralateral side against resistance to check for sternomastoid weakness
- The **hypoglossal nerve (XII)** is tested by asking the patient to stick out their tongue and move it from side to side. Look for wasting and weakness

At the end of the examination (if you have not already done so) you should ask to:

- Examine patient's glasses
- Look at fundi
- Assess pupillary responses to light and accommodation, offer to check corneal **(V)** reflexes

Suggested scheme for examination of the cranial nerves – infant

Don't panic! Observation is the key. Squint and facial nerve palsies would be the most likely scenarios in the neurology station. Babies are used far less frequently as the candidates do not get a uniform experience. You may be allowed to substitute questions for parts of the examination; making a baby cry to see if they have a facial palsy would not go down well! A modified cranial nerve examination is summarised below.

Cranial nerve(s)	Activity	Comment
I	Smell	At least mention
II	Visual acuity	Response to examination?
III	Eye movement	Can move up, down, in
IV	Eye movement	Can move down and out
V	Chewing	Ask parent if baby can chew
VI	Eye movement	Lateral
VII	Crying? Smiling?	Ask re: facial symmetry when crying
VIII	Hearing	Is there a startle reflex?
IX	Gag	Do not test gag in exam!
X	Phonation	Palatal movement
XI	Head turning	Equal both sides
XII	Tongue protrusion	Central?
V, VII, IX	Sucking	If absent, is there hypotonia?
IX, X, XI	Swallowing	Ask if intact

Table 11.1: Summary table of cranial nerve examination – infant

Notes on cranial nerve lesions

Origins

- III–IV: mid brain
- V–VIII: pons
- IX–XII: medulla or bulb

Olfactory nerve (I)

Traverses the cribriform plate to the frontal lobes, so lesions are most commonly detected in upper respiratory tract infections (URTIs), basal skull fractures, purulent meningitis, hydrocephalus. Remember anosmia also occurs in Kallman syndrome (associated hypogonadotrophic hypogonadism) caused by embryological neuronal migration defect.

Optic nerve (II)

Nerve fibres pass through the optic disc to the optic nerve. Nasal fibres decussate at the chiasm, and optic tracts carry impulses to the lateral geniculate bodies. The optic radiation carries the impulses to the visual cortex.

Patterns of visual defect

Patterns of visual field defect occur thus:

- **Blindness in one eye** – retinal, lens, optic nerve (chronic uveitis, optic nerve glioma, optic atrophy, septo-optic dysplasia)

- **Bitemporal hemianopia** – craniopharyngiomas, pituitary tumours affecting the chiasm
- **Homonymous hemianopia** – contralateral lesions posterior to the chiasm in the optic tract. This can be seen in cortical lesions such as cerebral palsy
- **Scotoma** – irregularly shaped visual field deficit arising from anywhere along the visual pathway; could be bilateral or monocular

Oculomotor nerve (III)

This nerve supplies the inferior, medial and superior rectus muscles as well as the inferior oblique muscle. Palsy is apparent as a dilated pupil that faces 'down and out'. Usually congenital, associated with chromosomal abnormalities or cerebral palsy. Acquired palsy may indicate tumours, infection or head injury.

Trochlear nerve (IV)

This nerve controls the superior oblique muscle and has a long intracranial course. Palsy usually presents subtly as head tilt towards the unaffected eye. On examination there is upward deviation of the eye and failure of downward and lateral gaze; with diplopia. Causes include raised intracranial pressure as the nerve has a long intracranial course. Will need surgical correction if head tilt is significant.

Trigeminal nerve (V)

Has three sensory divisions (ophthalmic, maxillary, mandibular), which pass into the trigeminal ganglion at the apex of the petrous temporal bone. The motor fibres originate in the pons and supply the muscle of mastication.

Lesions are caused by:

- Brainstem gliomas
- Infarction/AV malformation
- Acoustic neuromas
- Chronic suppurative otitis media
- Cavernous sinus thrombosis

Abducens nerve (VI)

The abducens nerve controls the lateral rectus muscle. Lesions of this nerve cause a marked convergent paralytic squint. The affected eye is turned nasally and is unable to abduct fully. This condition is rarely congenital. Acquired causes include raised intracranial pressure, tumours, arteriovenous (AV) malformations, meningitis.

Facial nerve (VII)

The facial nerve has a long intracranial course: initially it is closely related to the sixth nerve (lesions in the pons therefore tend to affect both nerves) and then it emerges from the cerebellopontine angle (lesions here also affect nerves V, VI and VIII) and runs in the petrous bone in close relation to the middle ear.

It is key to remember that:

An **Upper motor neurone** lesion causes weakness of the **lower part** of the face on the side opposite the lesion; **the forehead is spared** as it is bilaterally innervated.

A **Lower motor neurone** lesion causes weakness of the **whole side** of the face on the same side as the lesion.

Auditory nerve (VIII)

Passes through cerebellopontine angle.

Glossopharyngeal (IX), vagus (X) and hypoglossal (XII) nerves

All leave through the jugular foramen and are affected by lesions at the base of the skull such as achondroplasia and Arnold–Chiari malformation.

Facial nerve palsy

Examination

○ Smile
○ Frown
○ Raise the eyebrows (if this cannot be done, suspect a lower motor neurone lesion on that side)
○ Puff out the cheeks
○ Whistle
○ Close the eyes and keep them closed against a resistance
○ **Finally**, offer to check the blood pressure, look in the ears for signs of infection or rash, inspect the skin for a rash (?erythema migrans), assess hearing (?acoustic neuroma) and look for signs of exposure keratitis in the eyes (from incomplete lid closure)

Tip

Lower motor neurone lesion affects the upper and lower face
Upper motor neurone lesion spares upper face

Aetiology of facial nerve palsy

Lower motor neurone lesions:

- Bell's palsy (by far the commonest)
- Chronic serous otitis media
- Viral infections – mumps, Epstein–Barr virus
- Ramsay Hunt syndrome – an eruption of herpes zoster vesicles on the posterior wall of the ear canal accompanied by a lower motor neurone facial nerve palsy. Spontaneous recovery usually occurs
- Lyme disease
- Guillain–Barré syndrome (usually bilateral)
- Congenital
- Trauma – obstetric forceps, skull fractures
- Intracranial tumours, leukaemia

Upper motor neurone lesions:

- Cerebral palsy
- Tumours
- Moebius syndrome – loss of function of motor cranial nerves; child will have a strabismus and an immobile face because cranial nerves VI and VII most commonly affected. Extremely rare.

Bell's palsy

Usually unilateral; most are idiopathic.

Management

- Exclude hypertension
- Exclude other causes:
    - Lyme disease, particularly if bilateral. Many centres advocate borrelia serology testing for any patient with Bell's palsy who has visited a tick-infested area in the preceding few months
    - Varicella (from history, examination, +/– varicella titres)
    - Leukaemia (thorough examination and screen with FBC and film)
- Good eye care is very important – tape the eyes shut at night, and use artificial tears
- Treat with steroids if less than 7-day history
- Oral aciclovir has been used (usually in conjunction with steroids), but there is no clear evidence of benefit unless the child has Ramsay Hunt syndrome

Prognosis

- The prognosis is generally good and is better in children than in adults

- ❍ Most patients show signs of recovery by 3 weeks to 3 months. If lasts over 3 months, MRI should be considered

Muscular disorders causing facial weakness

- ❍ Myotonic dystrophy
- ❍ Facioscapulohumeral muscular dystrophy
- ❍ Congenital myopathies
- ❍ Mitochondrial myopathies
- ❍ Myasthenic syndromes

Bulbar and pseudo-bulbar palsy

A **bulbar palsy** describes a **lower motor neurone** lesion resulting in weakness of the muscles supplied by those cranial nerves whose nuclei lie in the medulla (or bulb), ie IX–XII. It is very unusual in children.

A **pseudo-bulbar palsy** results from bilateral supranuclear **upper motor neurone** lesions of the lower cranial nerves, resulting in poor tongue and pharynx movement. This is far more common than bulbar palsy, particularly in association with spastic quadriplegic cerebral palsy.

Signs of a pseudo-bulbar palsy

- ❍ Stiff spastic tongue that is not wasted
- ❍ ‘Dry’ voice and dysarthria
- ❍ Preserved gag and palatal reflexes
- ❍ Exaggerated jaw jerk

Suggested scheme for examination of the eyes

- ■ Look at the child for any obvious clues, eg squint, ptosis, dysmorphology
- ■ Does the child wear glasses?
- ■ Look at the eyes – conjunctiva, pupils, lids
- ■ Can the child see? Test both eyes separately; visual acuity
- ■ Visual fields
- ■ Eye movements
- ■ Check for nystagmus
- ■ Accommodation
- ■ Pupil responses – direct and consensual light reflexes
- ■ Cover test
- ■ Fundoscopy

Notes on eye examination

Observation

- This is very important, much pathology being visible on observation.
- Look first at the child:
  - Are there any obvious dysmorphic features?
  - Is the child thriving?
  - Is the child displaying behaviour suggestive of visual impairment, such as light gazing (cortical disease) or eccentric fixation with nystagmus?
  - With infants and very small children, try to achieve eye contact. See if the child will fix and follow in the horizontal and vertical planes. If a bright object is moved quickly from a central position to a peripheral one, you should see re-fixation on that object. This can only occur if the visual field on that side is intact.
- Look closely at the eye for the following:
  - Squint
  - Coloboma
  - Corneal clouding
  - Lesions of the iris
  - Ptosis
  - Anophthalmia (glass eye)
  - Microphthalmia.

The effect of glasses

This is clearly of importance and it is sensible to look at a few children with their glasses on before the exam to assess their vision and the effect of glasses.

Convex glasses correct long-sightedness (difficulty with near vision).

Concave glasses correct short-sightedness (difficulty with distant vision).

Visual acuity

For a **small child** the information you will gain will be limited and should be augmented by comments about their behaviour. Check that they can fix and follow in the horizontal and vertical planes. Use an interesting but silent toy or your face.

Tip +

Take care to keep quiet, because if you give auditory clues you will render the examination invalid (and disappoint the examiner!)

For **older children** use a suitable picture or story book (age dependent – don't ask a 4-year-old to read!) for near vision and use a Snellen chart for far vision. Test both eyes separately with and without glasses (if worn).

Tip +

Check vision in both eyes separately and don't get caught out by unilateral visual loss. If you suspect early on that a child is blind in one eye, continue to examine both eyes as instructed

Visual fields/eye movements/nystagmus

See 'Suggested scheme for examination of the cranial nerves – older child' (Section 11.2) and see Section 11.3 for nystagmus.

Accommodation

Ask the child to fix their gaze on your finger held in front of your face. Move your finger towards the child until it is touching their nose. Note their eye movements and pupillary responses.

Squint testing

See below.

Pupil responses

Test direct and consensual light reflexes.

Fundoscopy

Many candidates are asked to do this and you must appear proficient. Remember to examine for the red reflex first (from a distance of around 20 cm), then examine the anterior chamber and then the fundus. If the child is wearing glasses, remember to ask if they can remove them!

11.3 Cases and conditions: cranial nerves and eyes

Ptosis

This is a very common short case and you will be invited either to comment on or examine the child's eyes.

Instructions to candidate

Examine this 12-year-old boy with ptosis.

Case

> *This young man has a left ptosis. He is a well-looking boy and there are no dysmorphic features. I would like to examine his eyes. His eyes look normal. The pupils are symmetrical. There is no obvious squint. I would like to test his vision – candidate gets him to read – vision appears normal in both eyes (tested separately). His visual fields and eye movements are normal. His direct and consensual reflexes and his ability to accommodate are normal.*
>
> *I suspect therefore that his ptosis is congenital.*

Notes on ptosis

The examiner will expect you to examine the eyes carefully and in doing so, come up with a likely cause of the ptosis. The ptosis will become apparent on inspection. At this stage, on noting the ptosis, you need to have a clear differential diagnosis in your mind.

Ptosis may be broadly classified as syndromic or neurological in origin.

Most likely in exam:

- ❍ Horner syndrome
- ❍ Third nerve palsy
- ❍ Myasthenia gravis
- ❍ Congenital idiopathic ptosis

Full differential diagnosis of ptosis

Syndromic

- Noonan syndrome
- Rubinstein–Taybi (microcephaly, broad thumbs and toes, significant learning difficulties)
- Smith–Lemli–Opitz syndrome
- Marcus Gunn (aberrant innervation causes the affected lid to raise and wink when the child chews or cries)

Neurological

- Third nerve palsy
- Horner syndrome (see below)
- Myasthenia gravis
- Dystrophia myotonica
- Craniosynostosis
- Neuroblastoma
- Migraine

Others

- Congenital idiopathic cause (evaluate early to prevent amblyopia)
- Ocular tumours (rhabdomyosarcoma)

Notes on Horner syndrome

Lesion of the sympathetic nervous system in association with the brainstem or cervical cord or a lesion of the sympathetic plexus in association with the carotid artery.

Features are:

- Partial ptosis
- Pupil constriction
- Enophthalmos
- Ipsilateral anhidrosis
- Heterochromia iridis (complete when one iris is a different colour from the other; partial when part of one iris is a different colour from the rest) – indicates congenital Horner syndrome
- Normal direct and consensual reflex

Squint

Examination of a squint (see Box) is commonly asked for in the exam and needs to be practised carefully. It is best to get an orthoptist to help you to perfect your technique. Beware of epicanthic folds or hypertelorism giving a false impression of squint.

Examination of squint

○ Check the child can see first – with both eyes!
○ Corneal reflections
○ Eye movements
○ Cover test

Notes on squint examination

If you are told that a child has a squint, move straight on to testing after checking the acuity. If you notice a squint during a full eye examination, offer to test the child first without and then with their glasses. The child needs to be co-operative for this to work.

Corneal reflections

Observe both eyes while shining a point source of light at both corneas (corneal reflections). The light source should be held about 30 cm from the eyes and in different planes. If the eyes are straight, the reflection from both corneas will be symmetrical. If a squint is present, the reflection will be asymmetrical.

Eye movements

Eye movements should be examined in all directions using an interesting object. If the eyes deviate, ie there is asymmetry, then a squint is present.

If the deviation is worse in one direction, the squint is paralytic and is caused by a palsy of one of cranial nerves III, IV or VI (for the features of each lesion, see Box). Trauma and tumours are the commonest causes of acquired paralytic squint, and more than one cranial nerve may be affected.

If deviation is the same in all directions, the squint is non-paralytic.

Features of a third (oculomotor) nerve lesion

- ○ Complete ptosis
- ○ Diplopia
- ○ Downward and lateral gaze (unopposed lateral rectus and superior oblique muscles)
- ○ Pupil dilatation
- ○ Failure of the pupil to react to light or to accommodate

Features of a fourth (trochlear) nerve lesion

- ○ Diplopia
- ○ Upward deviation of the eye, +/− head tilt towards unaffected eye (often subtle)
- ○ Failure of downward and lateral gaze (failure of the superior oblique muscle) – difficult to read

Features of a sixth (abducens) nerve lesion

- ○ Diplopia
- ○ Unable to abduct on affected side (failure of the lateral rectus muscle), leading to a convergent paralytic squint

The following tests should be done at near and far vision, first with glasses and then without. This avoids missing a squint caused by excessive adduction secondary to hypermetropia.

Inward deviation is denoted by 'eso'.

Outward deviation is denoted by 'exo'.

Cover/uncover test

This examines for a manifest squint (tropia). You will note an asymmetrical corneal light reflection. Ask the child to fix on an interesting object straight ahead at 1 m and then 6 m. Cover the eye that appears to be fixing. If the other eye takes up fixation then there is a manifest squint present.

Alternate cover test

Again, ask the child to fixate on an interesting object at 1 m and then 6 m. Rapidly alternate covering/uncovering between left and right eyes. Look at each eye as it is just uncovered: if one moves to take up fixation, a latent squint (phoria) is present on that side.

Non-paralytic squint

Most squints will be **non-paralytic**. You may be asked about treatment of non-paralytic squint. There are two main aims. The first is to achieve the best possible vision in each eye. Any underlying defects such as cataracts or refractive errors are corrected, and amblyopia is prevented with occlusion therapy. Secondly, achieve the best possible ocular alignment.

Surgery is commonly required.

Nystagmus

Nystagmus describes involuntary, abnormal eye movements that are rhythmic and have either a slow and fast phase (jerky) or are slow in both directions (pendular). Most causes are pathological, but note that some children have nystagmus at the extremes of lateral gaze, which is physiological. This can be more pronounced when the child is tired.

Instructions to candidate

Look at this boy's eyes.

Case

> *Jack is a well-looking 10-year-old boy who has pale skin, blue eyes and fair hair. Examination of his eyes reveals that he usually wears sunglasses, there is nystagmus and reduced visual acuity. Red reflexes are easily visible. I suspect he has oculocutaneous albinism.*

What is the inheritance?

○ Autosomal recessive (ocular albinism alone is X-linked recessive)

Notes on oculocutaneous albinism

- Commonest form of albinism; there are four different subtypes
- In all types there is an absolute or relative lack of melanin pigment in eyes, hair and skin
- Opthalmological problems include photophobia, strabismus, reduced acuity as well as nystagmus
- The principal long-term consequences are blindness and skin cancer. Avoidance of sunlight, where practicable, and use of high-factor sunscreens, are essential
- A differential diagnosis of oculocutaneous albinism is Chediak–Higashi syndrome, a rare condition in which there is increased susceptibility to bacterial infections as well as hypopigmented eyes, skin and hair.

Notes on nystagmus

- **Cerebellar nystagmus** is the most important type to recognise. It is jerky and is usually horizontal, and worsens on looking to the side of the lesion. The fast (return) component is directed towards the side of the lesion
- **Vestibular nystagmus** differs in that the slow phase is directed towards the side of the lesion
- **Vertical nystagmus** is usually due to a lesion of the brainstem at the pontomedullary junction (roughly at the foramen magnum), eg achondroplasia, or Arnold–Chiari malformations. It can also be caused by phenytoin or carbamazepine toxicity.
- **Ocular nystagmus** with slow, searching movements occurs in blindness
- **Congenital nystagmus** is present at birth, although it is often not fully recognised until after the first few months. It is usually bilateral and horizontal. If it is idiopathic it often improves with age, although it is important to be aware that there are some more serious causes which carry a poorer prognosis

Fundoscopic findings – notes on various signs

Papilloedema

The stages of papilloedema are:

1. The optic nerve becomes hyperaemic
2. Small capillaries on the optic disc disappear as they become compressed
3. Venous pulsations are no longer seen as the intracranial pressure increases
4. The border of the optic disc becomes indistinct
5. Flame-shaped haemorrhage appears around optic disc

Florid papilloedema is a late sign of raised intracranial pressure.

Causes

- Raised intracranial pressure
- Brain tumours – craniopharyngioma, brainstem glioma, astrocytoma, medulloblastoma
- Leukaemia
- Idiopathic intracranial hypertension (IIH)
- Hydrocephalus
- Systemic hypertension
- CNS infection
- Encephalopathy
- Intracranial haemorrhage
- Craniosynostosis

Cherry red spot – causes

This occurs in certain lysosomal storage diseases.

Seen at the macula, this is the comparatively reddened appearance when one can see through the fovea (which does not build up storage material as relatively less cells) through to reddened choroid below.

This area will be 'cherry red' as it is contrasted with the surrounding retina, which will be pale with build-up of storage material.

Such conditions are:

- Tay–Sachs' disease
- Niemann–Pick's disease
- GM1 gangliosidosis
- Sandhoff's disease
- Mucolipidosis

Lens dislocation – causes

- Marfan syndrome – lens dislocation is present in 80% of adults and 50% of 5 year olds with Marfan syndrome. It usually dislocates superiorly and temporally. Symptoms include blurred vision (due to refractive changes) and diplopia. If the pupil is dilated, the edge of the lens may be seen as a black crescent on ophthalmoscopic examination. A clearer view is obtained with slit lamp examination; mention this in the exam
- Homocystinuria – lens dislocates downwards and nasally
- Ehlers–Danlos syndrome

Cataracts – causes

- Metabolic disease, eg galactosaemia

- Wilson's disease
- Hypoparathyroidism
- Diabetes
- Dystrophica myotonica
- Intrauterine infections
- Traumatic insult to the eye

Corneal clouding – causes

- GM1 gangliosidosis
- Fucosidosis
- Mucopolysaccharidosis
- Mucolipidosis

Systemic conditions with eye signs

Condition	Sign
Neurofibromatosis	Lisch nodule, ptosis
Wilson's disease	Kayser–Fleischer ring
Ataxia telangiectasia	Conjunctival telangiectasia
Down syndrome	Brushfield spots – not pathognomonic
Hyperthyroidism	Exophthalmos, lid lag
Juvenile idiopathic arthritis	Iridocyclitis

Table 11.2: Systemic conditions with eye signs

Miscellaneous eye problems

Coloboma

Sometimes seen in the examination. Either inherited (autosomal dominant) or as part of the conditions listed below:

- Aniridia–Wilms' association
- CHARGE association
- Goldenhar syndrome

Duane syndrome

A congenital abnormality of innervation causing simultaneous contraction of medial and lateral recti on attempted adduction of the affected eye. On examination they may have exotropia or esotropia, and on testing eye movements they may have failure to abduct or adduct the affected eye.

Moebius syndrome

This is manifest as a (usually) bilateral but asymmetrical lower motor neurone facial nerve palsy, associated particularly with sixth nerve palsies. These cause failure of abduction and paralytic convergent squints of the affected eyes. The cause is unknown, but as both cranial nerves are closely related it may be due to underdevelopment of the cranial nerve nuclei.

Uveitis

This is inflammation of the uveal tract consisting of the inner vascular coat of the eye; the iris, the ciliary body and choroid. Iritis may occur alone or in conjunction with the ciliary body as iridocyclitis. This commonly occurs insidiously without symptoms.

Children with seropositive (ANA) arthritis are in a high-risk group for uveitis and need to have regular eye reviews.

Case

Jane is a well-looking 8-year-old girl. She appears cushingoid. Examination of her eyes reveals conjunctival injection. The visual acuity of her left eye is markedly reduced. It was not possible to examine the visual fields. The pupil responses are present but slow on the left. Ophthalmoscopy reveals clouding of the cornea on the left. There is normal visual acuity in her right eye with normal pupil responses and no clouding of the cornea.

This would fit with chronic uveitis. I would like to proceed to a full systemic examination, paying particular attention to the joints and looking for the side-effects of steroids.

11.4 Central and peripheral nervous system

Suggested scheme for examination of the central and peripheral motor system

This needs to be done carefully and correctly. It is essential to practice and make your technique second nature. Remember to look for obvious clues first (eg obvious neurological deficit such as cerebral palsy, orthoses, wheelchairs). The examiner may limit you to examination of the arms or the legs. You should have a different approach to infants compared to that used with older children.

Infants

Most information is gained by **observation** and so spend some time watching and playing with the infant. Have a look at the child from head to toe when you walk into the room:

- Look for any obvious dysmorphology
- Try to get eye contact – see if the child will fix and follow in the horizontal and vertical planes
- Is the child moving any limbs? Is movement symmetrical?
- Remember that a baby lifting their right arm has at least grade 3 power in that limb. Ask the mother about hand preference, which is abnormal under 1 year
- Look specifically for leg scissoring (spastic diplegia), paucity of movement on one side (hemiplegia), overall paucity of movement (spinal muscular atrophy)

Perform a brief **neurodevelopmental assessment**. It is essential to know the normal milestones (see Section 13.2). During this assessment you will be able to assess **tone** and **power**:

- Pull the child to sit – use one hand to pull up both hands and the other to support the head. Is there any head lag? This is the best way to assess head control because if the child is held prone and the head is extended, this may reflect extensor spasm
- Hold the child by the trunk in the sitting position
- Assess standing posture if possible
- Hold the child up prone looking at tone and head control
- Place the child on the bed prone to see if the child supports their head, supports themselves on forearms (3 months), or supports themselves on outstretched hands (6 months)
- Proceed with further examination dependent upon the initial findings, eg tone and power in all four limbs
- Proceed to examine the **tendon reflexes** as you would in an older child. Remember to examine for **ankle clonus** (one or two beats are normal under 1 year) and **plantar reflexes** (up-going until 1 year).

Finally, examine for the presence of the primitive reflexes (eg Moro reflex, asymmetric tonic neck reflex, grasp, etc). These are described in Chapter 13. Give some warning before attempting a Moro reflex, as it is sometimes unpleasant for the infant. It may be sufficient to explain the procedure and then ask the parent whether you can perform it.

Older children

This should be done more conventionally, as most older children can co-operate with a more formal neurological examination. Observe the child as you walk in and introduce yourself. Ask the child to walk first unless there is an obvious reason why they cannot (eg because they are in a wheelchair). Look for clues as above. Observation of gait may tell you where to focus the rest of the examination:

- Look at muscle bulk and symmetry
- Inspect for scars (eg tendo-achilles shortening)
- Examine tone
- Examine power
- Examine the reflexes, including testing for clonus and plantar reflexes
- Examine co-ordination
- Further examination as appropriate (eg sensory, cerebellar, etc)

You may well be asked to focus on just a part of the neurological examination, such as a cerebellar assessment.

Upper motor neurone or lower motor neurone lesion?

This relatively straightforward piece of information (Table 11.3) is often asked for and is a reproducible discriminator between a good and bad candidate. It is very important to be ready for this question with the correct answer as a number of candidates when asked this do not appreciate what information the examiner is requesting.

Observe for clues then examine tone, power and reflexes.

Characteristics of an upper motor neurone lesion	▪ Increased tone ▪ Clonus ▪ Reduced power ▪ Increased reflexes
Characteristics of a lower motor neurone lesion	▪ Wasting ▪ Fasciculation ▪ Reduced tone ▪ Reduced power ▪ Reduced reflexes

Table 11.3: Characteristics of upper and lower motor neurone lesions

Tell the examiner which type of lesion this is and clearly explain how you came to this conclusion.

Suggested scheme for examination of gait

The examination of gait is very important and often asked of the candidate. The question can either be a direct one or be phrased indirectly such as **'examine this child's legs'**, when, providing the child can walk, gait examination is the appropriate first step.

Gait problems can be due to the following:

- Hemiplegia
- Diplegia
- Ataxia
- Neuromuscular disease
- Orthopaedic problems (can develop from spasticity)
- Rheumatological disorders (more likely to be in the 'musculoskeletal/other' station)
- Secondary to a limp

Children with abnormal gaits need to be seen (try your local child development unit or general outpatients) in the run-up to the exam. You will then have seen the commoner gait abnormalities and will be able to describe them comfortably under pressure. It is sensible to use a scheme for examination and one is suggested below. Before you launch into an examination, first check with the child or parents that the child can walk:

- Look at the child. Do they look dysmorphic? Is there an obvious hemiplegia or diplegia? Is there a built-up shoe?
- Look at their surroundings for equipment – are there walking sticks or a wheelchair nearby?
- Ask them to walk and decide whether their gait is spastic, ataxic or due to a neuromuscular problem. These categories are your best bet if you are in the neurology station!
- Don't forget to **look at the spine** at some point in the examination. It is an essential part of examining the lower limbs and candidates have failed for missing it out

Spastic gait

Ask the child to walk unaided – look at arm and leg movement for signs of a hemiplegia. The arm will be flexed on the affected side. If you suspect hemiplegia, ask the child to walk fast or walk on tip toe, which will make the neurology more pronounced. Continue looking – are the legs stiff and adducted, if so, a diplegia is likely. A diplegia will also become more pronounced if you ask the child to walk fast or on tip toe:

- If the child has either a hemiplegia or a diplegia you can then take the child to the examination couch and confirm the neurology with a careful peripheral neurological examination
- You may then be asked if you want to examine anywhere else – upper limbs, cranial (neurocutaneous syndrome)

Ataxic gait

If the gait is not spastic and seems unsteady and/or broad based, the next thing to consider is whether it is ataxic. Ataxia is the incoordination that results from either **sensory loss** or **cerebellar dysfunction.** In children the latter is more likely:

- Ask the child to walk heel-to-toe; you will need to demonstrate this. If the child is ataxic this will be difficult
- Then ask the child to stand still – they should fall over towards the side of a cerebellar lesion
- Ask the child to close their eyes. If the lesion is cerebellar there should be no deterioration. If the ataxia is a consequence of a dorsal column problem, the child will fall (Romberg's sign). The latter is rare in childhood
- If you suspect that the gait is ataxic and due to a cerebellar lesion, confirm this with a cerebellar examination. This is detailed below in the 'Scheme for cerebellar examination'

Neuromuscular problem

If neither of the above seem likely, the next possibility is that you are dealing with either a neuromuscular or a lower motor neurone problem:

- Look for a waddling gait and foot drop
- Examine the child for Gower's sign. Ideally, ask them to lie on the floor (if it is clean!) in a supine position and then to stand up. A positive Gower's sign occurs when the child has to turn from supine to prone (an important feature of the examination), then rises from the floor by pushing off and crawling up their legs with their hands. This means that they are trying to overcome proximal pelvic weakness. If it is not practicable for the child to lie on the floor, conduct the assessment from a sitting position, but tell the examiner that children with subtle weakness may not always have to crawl up their legs with their hands
- If the Gower's sign is positive, a peripheral neurological examination should be carried out and the back checked for a scoliosis or spina bifida

Others

Finally, consider either an orthopaedic or a rheumatological problem. Comment on whether the gait looks painful (antalgic).

Notes on examination of gait

Limp

You may see a limping child in this station or in the MSK station.

The aetiology of limp can be broadly divided into two groups: non-painful and painful. A child with a non-painful limp has a Trendelenburg gait. Here the child shifts their centre of gravity *over the affected side* for balance, and this is an indicator of proximal muscle weakness (by far the most likely cause in this station) or hip instability. The affected hip is *lower* than the unaffected hip.

Causes are:

- Neurological (muscular dystrophy)
- Congenital hip dislocation
- Slipped capital femoral epiphysis

A painful limp gives the child an antalgic gait. This gait is more likely to be seen in the Musculoskeletal/Other station. Here the child spends the least amount of time possible on the affected limb. This is Trendelenburg negative, which implies that the affected hip is *higher* than the unaffected hip, as the centre of gravity is shifted *away* from it.

Causes of a painful limp are:

- Infection
- Trauma
- Perthes' disease
- Rheumatological (juvenile idiopathic arthritis)

Scheme for sensory examination

This would be very difficult in the context of the exam. The appropriate equipment should be provided. Your approach to the child will be important and you need to encourage co-operation to gain maximum information. For older children you should have a polished technique so that you not only put the child at ease, but are able to concentrate on the signs elicited. The way in which you phrase your questions and the precise instructions that you give are key. When preparing for this station, ask a colleague or senior to watch you performing a sensory examination and give you feedback. You should test the following:

- Pain, light touch and temperature – spinothalamic tracts
- Joint position sense, vibration and proprioception – posterior columns

You may then be asked to give a sensory level if appropriate. This will require a thorough knowledge of dermatomes.

Scheme for cerebellar examination

If asked to examine cerebellar function you need to have your own scheme. Observation is important – stop and look first. During the examination, make sure that you do not move the child around more than is necessary.

Below is a suggested scheme:

- Introduce yourself and ask the child a question. Listen for dysarthria. Is there staccato speech?

- Think of the causes of ataxia while observing the child and look for associated signs, eg telangiectasia or pes cavus
- Ask the child to walk. Observe the gait, which will be ataxic and broad based, leaning towards the side of the lesion if unilateral
- Ask the child to sit to check for truncal ataxia
- Examine the arms and legs for hypotonia and hyporeflexia
- Test eye movements and look for nystagmus – horizontal and maximal, looking to the side of the lesion
- Test co-ordination. Ask the child to touch their finger and then your nose with their index finger. Look for past-pointing or intention tremor. Assess rapidly alternating movements (dysdiadochokinesis)
- Stand the child up and examine for **Romberg's sign** to differentiate sensory posterior column disease from cerebellar disease. The child is asked to stand with their feet just sufficiently apart to become steady. The child then closes their eyes. If the child has a cerebellar problem there is no deterioration in balance. If the child has a sensory ataxia then he or she will fall (Romberg's sign)

Abnormalities of posture and movement

During your neurological examination you may notice sustained abnormalities in the child's movement or posture. These abnormalities can usually be categorised as **tremor, myotonia, dystonia, chorea, athetosis or tics.**

Remember that dystonia and choreoathetosis are abnormalities of posture and movement secondary to extrapyramidal dysfunction.

Tremor

- ○ Involuntary, rhythmic trembling or shaking movement:
    - Physiological
    - Drug induced – salbutamol, thyroxine
    - Benign essential tremor – positive family history, intentional
    - Cerebellar – not present at rest (intentional)
    - Parkinson's (rare) – present at rest (not intentional)

Myotonia

- ○ The failure of muscle to relax after contraction:
    - Congenital myotonic dystrophy

Dystonia

- ○ Abnormal posturing of the limbs, trunk and face
- ○ Can occur in association with choreoathetosis:
    - Cerebral palsy, drug induced, Wilson's disease

Chorea

- Involuntary, rapid and irregular repetitive jerking. Either generalised or affecting one part of the body. Many potential causes including:
    - Cerebral palsy, drug induced, Wilson's disease, metabolic, autoimmune, infectious eg Lyme disease

Athetosis

- Involuntary, slow writhing movement of the limbs. Can occur with chorea (choreoathetosis):
    - Athetoid cerebral palsy

Tic

- A sudden, brief and purposeless stereotyped movement which can (at least to some extent) be suppressed by voluntary effort
- Can be motor, eg facial twitching, shrugging of shoulders
- Vocal tics can occur, such as grunting
- Most are functional, and there can be a genetic tendency (ask to take a family history)
- Can also be seen in cerebral palsy, autoimmune (after a streptococcal infection) or as a side-effect of dexamphetamine in children being treated for attention deficit hyperactivity disorder (ADHD)

When you have picked up signs of disordered movement or posture, present your findings succinctly to the examiner as in the case below.

Case

John is a 12-year-old boy in a wheelchair. He is mentally alert but has dystonic posturing with choreoathetoid movements. The diagnosis is cerebral palsy, and possible causes of the movement disorder include a neonatal encephalopathy or kernicterus.

Useful lists

These lists are essential knowledge and will enhance the way you present an abnormality. For example, an examiner may disagree with your grading of power and ask you how you derived it.

Power – MRC grading

0 No contraction
1 Flicker or trace of contraction
2 Active movement with gravity eliminated
3 Active movement against gravity
4 Active movement against gravity and resistance
5 Normal power

Reflexes – spinal segments and nerve roots involved

Reflex	Spinal segment	Nerve
Biceps	C5,6	Musculocutaneous
Supinator or brachioradialis jerk	C5,6	Radial
Triceps	C6,7	Radial
Superficial abdominal	T7–12	
Knee	L3–4	Femoral
Ankle	S1	Tibial
Plantars	S1	

Table 11.4: Reflexes – the spinal segments and nerve roots involved

Spinal levels of some cutaneous reflexes

- Superficial abdominal: T7–12
- Cremasteric: L1
- Plantar: S1
- Anal: S4–5

Myotomes

Movement tested	Nerve root
Shoulder abduction	C5/6
Elbow flexion	C5/6
Elbow extension	C7/8
Finger flexion	C8
Finger abduction	C8/T1
Hip flexion	L1
Hip adduction	L2
Knee extension	L3
Foot dorsiflexion	L4
Foot plantar flexion	S1
Knee flexion	S2
Hip flexion	S3

Table 11.5: Myotomes

11.5 Cases and conditions: central and peripheral nervous system

Cerebral palsy

Children with varying types and severities of cerebral palsy can be seen. The cases used as examples are meant to get you thinking about how to deal with and talk about children with cerebral palsy who you see in the exam. You need to make contact with whoever runs the child development unit in your area and arrange to see and discuss some children with cerebral palsy.

Remember the definition – the cerebral palsies are a group of persistent, but not necessarily unchanging, disorders of movement and posture due to a non-progressive disorder of the developing brain. As well as problems with motor development, there are often problems with visual, auditory, sensory, social and cognitive development.

As this excludes diagnoses such as brain tumours and spinal cord disease, try not to mention them as causes.

Children with cerebral palsy and their families are also often asked to help out in the history taking and management planning station and a complex case is discussed in the relevant section, along with extensive notes on this condition (page 41 onwards).

When interacting with a child who has cerebral palsy do not make the assumption that there are learning difficulties. Many children have a cognitive ability that exceeds their ability to communicate. Always talk to patients as well as parents, even if this is difficult and there is little reciprocation – after all this should be normal practice on the wards and in outpatient clinics.

Cases

For the clinical station, most patients with cerebral palsy can be categorised into those with:

○ Hemiplegia
○ Monoplegia, diplegia or quadriplegia
○ Ataxic cerebral palsy
○ Dyskinetic (eg athetoid) cerebral palsy

NB Some children have a mixed form of cerebral palsy.

Case one

Peter is a thin 5-year-old boy who walks with a Trendelenburg-positive gait; there is muscle wasting and decreased tone in the left leg. He has brisk reflexes with up-going plantars and short tendo-achilles in the left side. There is no involvement of the left arm or any other limb.

*The **diagnosis** is a **monoplegia** involving the left leg.*

Discussion points

- ○ You may be asked to go through causes. Remember to categorise them as prenatal, perinatal or postnatal and to appreciate that the cause may well be unknown
- ○ Why has he got a tight tendo-achilles on the affected side (contractures)?
- ○ How would you treat his shortened Achilles tendon (physiotherapy, surgery if necessary)?

Case two

Jade is a thin-looking 10-year-old girl with gross muscle wasting of all four limbs. She is lying in a windswept position (alternatively, she could be in a wheelchair with support). She is drooling. There is generalised hypertonia with contractures and increased reflexes. Both plantars are up-going ...

*The **diagnosis** is **spastic quadriplegia**. I would like to examine the cranial nerves as she may have a pseudo-bulbar palsy.*

Discussion points

- ○ Why might there be problems feeding her orally (pseudo-bulbar palsy)?
- ○ What alternative methods of feeding could be used (gastrostomy)?

Case three

Ellie is a thin 9-year-old girl who is sitting supported in a wheelchair. She is drooling. There is truncal hypotonia with four-limb hypertonia and hyper-reflexia with striking choreoathetoid movements ...

*The **diagnosis** is **athetoid cerebral palsy**.*

Discussion points

- This type of cerebral palsy falls under the group of dyskinetic forms, which result from a lesion in the extrapyramidal system or basal ganglia. In dyskinesia there is increased muscle tone and abnormal posturing, as well as involuntary movements. These are more obvious when the child is attempting an activity voluntarily.
- If you are asked about possible causes, a neonatal encephalopathy (formerly called a 'hypoxic-ischaemic encephalopathy'), could have led to this picture. Another neonatal problem that may have been the cause is kernicterus secondary to severe unconjugated hyperbilirubinaemia.

Case four

> *Robert is a well-nourished 14-year-old boy with a broad-based ataxic gait. He has increased tone and brisk reflexes in his legs, with normal tone and reflexes in his arms. He has nystagmus and an intention tremor with past-pointing. He is Romberg's negative.*
>
> *I think he has a* ***cerebellar ataxia with a diplegia,*** *and the diagnosis is ataxic cerebral palsy.*

Discussion points

- This would be a difficult case and a good differential of the causes of cerebellar ataxia would be called for
- The important thing is to demonstrate good examination technique and have confidence in your findings, even if you have difficulty pulling them together to make a diagnosis

Neuromuscular disease

This is another group of disorders seen with disproportionately high frequency in the exam and hence you need to know about them in considerable detail. These include diseases that affect the lower motor neurone at one of the following sites: **anterior horn cell, nerve fibre, neuromuscular junction** or **muscle.**

There are two main scenarios, which we will deal with separately: the older child with neuromuscular disease and the floppy infant.

The older child

Commonly seen cases in the exam:

- Duchenne muscular dystrophy
- Spinal muscular atrophy type III (Kugelberg–Welander's disease)

- ❍ Dermatomyositis
- ❍ Peroneal muscular atrophy

Differential diagnosis of neuromuscular disease in the older child (see Table 11.6).

This may well be asked for once you have presented your findings. The easiest way to remember this for most people is to think of the reflex arc and start 'proximally' at the anterior horn cell and move distally from the nerve fibre to the neuromuscular junction and then muscle. More likely diagnoses in the exam are listed first in each section.

Anterior horn cell	Spinal muscular atrophy
	Poliomyelitis
Nerve fibre	Peroneal muscular atrophy (Charcot–Marie–Tooth)
	Infectious polyneuritis – Guillain–Barré syndrome
	Leukodystrophies
	Poisons
Neuromuscular junction	Myasthenia gravis
Muscle	Muscular dystrophy – Duchenne – Becker – Facioscapulohumeral dystrophy – Limb girdle dystrophy
	Dystrophia myotonica
	Inflammatory, eg polymyositis, dermatomyositis
	Metabolic disease, eg glycogen storage disease
	Thyroid disease
	Steroids

Table 11.6: Differential diagnosis of neuromuscular disease in the older child

Neuromuscular disease in the older child: cases

Case one

Andrew is a thin-looking 14-year-old boy who walks with a waddling gait. He has a positive Gower's sign. Examination of his peripheral nervous system shows reduced muscle bulk, hypotonia and absent tendon reflexes. There is a proximal muscle weakness in all four limbs. When his arms are outstretched, he has winging of the scapula and a fine tremor.

The diagnosis is spinal muscular atrophy type III (Kugelberg–Welander's disease) which affects the anterior horn cells.

Notes on spinal muscular atrophy type III

- Autosomal recessive and gene deletion known – diagnosis possible from DNA
- Presents at age 5–15 years with waddling gait and proximal muscle weakness, which often manifests itself when the patient is trying to climb the stairs
- Gower's positive
- Hypotonia
- Reduced power – mostly proximal
- Reduced reflexes
- Tongue fasciculation

Differential diagnosis

Limb girdle muscular dystrophy – see 'Notes on other muscular dystrophies' below. The two conditions have similar clinical features and it is difficult to distinguish between them. This needs DNA analysis, electromyography and nerve conduction studies.

Case two

Instructions to candidate:

Examine this girl's legs.

Jennifer is a well-looking 10-year-old girl who is lying flat in bed. She has reduced tone in her lower limbs and symmetrical weakness, which is more pronounced distally than proximally. She has absent knee and ankle jerks.

*The diagnosis is **Guillain–Barré syndrome**; I would also like to examine her cranial nerves, perform spirometry and check her blood pressure.*

Notes on Guillain–Barré syndrome

This acute inflammatory demyelinating polyneuropathy usually starts 10–14 days after a viral prodromal illness. Although it is rare, you may see a case if there is a recovering patient on the ward:

- ○ It is characterised by an ascending flaccid paralysis. Classically symmetrical; there may be a degree of minor asymmetry
- ○ Distal sensory loss or paraesthesia occur but are less marked than the motor symptoms and signs
- ○ Neuropathic pain is not uncommon
- ○ On examination there is reduced tone and absent or reduced tendon reflexes
- ○ There will be a flexor plantar response
- ○ Examine the cranial nerves, as these patients are prone to ptosis and seventh nerve palsies

The diagnosis is clinical, and children with a suspected case will undergo supporting tests including MRI of the brain and spine, lumbar puncture (high CSF protein) and nerve conduction studies. The illness must be monitored with daily spirometry, as the onset of respiratory failure is insidious and life-threatening. Some patients require ventilatory support. Those with severe disease can have autonomic involvement with blood pressure instability and arrhythmias contributing to morbidity and sometimes mortality. All patients should have a baseline ECG on diagnosis.

Treatment in severe cases is with iv immunoglobulin or even plasmapheresis. Physiotherapy and rehabilitation are very important, so it is essential to mention them in the exam.

If you are asked about the prognosis, the main indicator is time to initial improvement after maximal weakness, with those who do not start to recover until more than 2 weeks after the start of the illness having the worst prognosis.

NB Patients who have had polio may (very rarely) be seen in the examination. Remember that polio starts with an influenza-type illness followed by muscle pain and then progressive ascending asymmetrical paralysis, which is worse in the legs.

Case three

Instructions to candidate

Look at this child's legs and in particular his feet.

> *Jay is a 12-year-old boy who has pes caves. There is associated weakness and wasting of the distal muscles of the leg, in particular he has weakness of foot dorsiflexion. There are absent knee and ankle jerks on both sides. His plantar response is down-going. I would like to test the sensation in his feet.*
>
> *The diagnosis is **peroneal muscular atrophy.***

Notes on peroneal muscular atrophy

- ○ A disorder of the nerve fibres – also known as hereditary motor and sensory neuropathy (HMSN) type 1, or Charcot–Marie–Tooth Disease. Usually manifests in late childhood. The most common form (HMSN 1A) is autosomal dominant. The peroneal and tibial nerves are damaged, causing foot drop and the characteristic distal wasting
- ○ Sensory nerves conveying proprioception and vibration also become involved
- ○ Treatment of the foot drop is by stabilisation of the ankle joint with ankle–foot orthoses

The main differential diagnosis is Friedreich's ataxia, which is characterised by ataxia, Romberg's positivity, absent tendon reflexes and up-going plantars.

Case four

Samuel is an 8-year-old boy who is able to walk with the aid of a walking frame and foot orthoses. He has a wide-based gait and an exaggerated lumbar lordosis. His calves are hypertrophied. On examination of the peripheral nervous system it is apparent he has a positive Gower's sign and reduced muscle bulk of his thighs; there is weakness of his hip flexors. There are absent knee jerks but brisk ankle jerks. I would like to examine his back for a scoliosis.

The above findings suggest a muscular disorder. The diagnosis is ***Duchenne muscular dystrophy****.*

Questions

- How do you confirm the diagnosis?
- What is the differential diagnosis?
- What is the prognosis?

Notes on Duchenne muscular dystrophy (DMD)

- Commonest disabling neuromuscular disorder of childhood. X-linked recessive inheritance with an incidence 1 in 3500 live-born boys. The mutations are in the dystrophin gene. The gene locus is at Xp21. A third of cases represent a new mutation. Girls may be symptomatic as a consequence of the random inactivation of one of the X chromosomes.
- Usually presents between the ages of 2 and 5 with delayed walking, waddling gait, tip-toe walking, excessive falls and difficulty with stairs. Calf muscle hypertrophy is often present. Most reflexes disappear early in the disease, apart from the ankle jerk which disappears late. A variable degree of cognitive impairment is often present and speech delay is often seen in tandem with or before the delayed walking.
- Creatinine phosphokinase is usually high at birth and at diagnosis.
- Diagnosis is by DNA studies looking at the dystrophin gene. Electromyography (EMG) and muscle biopsy for histochemical studies can also be done, particularly if initial DNA studies are inconclusive. Both show typical features. Do mention genetic counselling of the family as part of the management after confirmation of this disease.
- Complications arise from cardiac, respiratory and skeletal muscle involvement; DMD should be described as being a multisystem disorder. Dilated cardiomyopathy can be a significant problem as the child enters adulthood, with left ventricular failure and arrhythmias often needing medical intervention.

- Scoliosis is common and is seen in around 80% of these children; it exacerbates their pre-existing restrictive lung disease. Spinal fusion surgery is often needed. Physiotherapy input is important.
- These children lose their ability to walk unaided between 7 and 12 years of age. A walking frame is used first, followed by a wheelchair as the child enters their second decade.
- Steroids are currently the only pharmacological treatment. They can slow the rate of deterioration in muscle strength and have some positive effects on respiratory function. Side-effects need to be closely monitored.
- Physiotherapists and occupational therapists play a key role in the multidisciplinary team.
- Although non-invasive ventilation can increase life expectancy by a few years, this condition is usually fatal during the second or third decade, with respiratory failure being the commonest cause of death.

Notes on other muscular dystrophies

Becker muscular dystrophy

- Incidence is 1 in 30 000. Presents later than Duchenne muscular dystrophy. The gene defect is at the same locus as Duchenne muscular dystrophy. Cardiac and respiratory muscle involvement is rare. The creatinine phosphokinase is high at diagnosis. EMG and muscle biopsy are helpful in establishing a diagnosis.

Facioscapulohumeral muscular dystrophy

- Autosomal dominant. Facial and shoulder girdle muscles are affected. The face is typically expressionless and there is also winging of the scapula, with shoulder muscle weakness.

Limb girdle muscular dystrophy

- Most cases are autosomal recessive. Onset is in late childhood. Hip and shoulder muscles become atrophied and weak, and the condition progresses distally. Toe walking and a waddling gait are seen as the illness progresses, and most patients are wheelchair bound by the age of 30 years.
- Note that affected children can develop calf muscle hypertrophy and ankle contractures, which causes confusion with Becker muscular dystrophy.

Tip +

Another condition that can present with muscle weakness is **dermatomyositis**, which is covered in Section 12.1. Asymmetry helps to distinguish it from other muscle disease, eg limb girdle dystrophy

The floppy infant

You may be asked to examine an infant in whom it is apparent that there is poor truncal tone, and as your examination progresses you realise that you are dealing with a 'floppy' infant. There will be head lag, and as you observe and examine the infant you should ask yourself whether they are just floppy (which implies a central cause), or whether they are floppy and weak (which suggests that the cause is more likely to be peripheral, ie peripheral nerve, muscular or neuromuscular junction).

However, be aware that infants with cerebral palsy (central cause) can be floppy and weak.

Commonly seen cases in the exam:

1. Spinal muscular atrophy type I (Werdnig–Hoffman's disease) – a floppy and weak infant *with absent reflexes* should make you think of this
2. Myotonic dystrophy
3. Down syndrome

Differential diagnosis

If you see a hypotonic or floppy infant, you may be asked for a full differential diagnosis. It is important to remember that there are many causes of the 'floppy infant' that can be seen in the exam. Both neurological (helpful to split into central or peripheral if possible) and systemic conditions need to be considered. A full systematically organised differential diagnosis is listed (Table 11.7). Try to organise your differential diagnosis in a similar way to the approach of neuromuscular disease seen previously.

Remember that if you are allowed to ask questions, a pregnancy/birth and neonatal history (especially asking whether there were any features of a neonatal encephalopathy) may give clues as to the cause. Always ask about development; children with a central cause for their hypotonia are more likely to have learning and social difficulties +/– epilepsy.

Central	Encephalopathy
	Cerebral palsy
	Intracranial haemorrhage
	Degenerative disease, eg infantile Gaucher's disease
	Neurometabolic disease, eg Zellweger's, previous significant hypoglycaemia
Spinal cord	Spina bifida
	Transection of the cord, eg following complicated breech delivery
	Haematoma
	Tumour
Anterior horn cell	Spinal muscular atrophy type I (Werdnig–Hoffman's disease)
	Poliomyelitis
Nerve fibre	Demyelinating disease, eg Guillain–Barré syndrome
Neuromuscular junction	Transient neonatal myasthenia gravis, congenital myaesthenia
	Botulism
Muscle	Congenital myopathies
	Congenital myotonic dystrophy
	Congenital muscular dystrophies
	Pompes' disease
Other causes of hypotonia	Down syndrome
	Prader–Willi syndrome
	Acute and chronic childhood illnesses Hypothyroidism Hypercalcaemia Renal tubular acidosis Rickets Coeliac disease Cystic fibrosis

Table 11.7: Differential diagnosis for floppy infant

Neuromuscular disease in the floppy infant: cases

Anterior horn cell

Case

> *Samantha is a happy-looking 6-month-old child who is visually alert. She is lying flat on her back with obvious paucity of movement and only minimal antigravity movement. She has a bell-shaped chest and paradoxical 'see-saw' respiration. When pulled to sit and on ventral suspension there is head lag and poor tone. She has no sitting posture. Tendon reflexes are absent.*
>
> *The diagnosis is infantile spinal muscular atrophy type I (Werdnig–Hoffman's disease)*

Different mutations in the gene for spinal muscular atrophy produce different illnesses in terms of age of onset and rate of progression. Type I is the most severe.

Discussion points

- Investigation and differential diagnosis – requires neurology referral, EMG/ nerve conduction studies, and genetic counselling (gene probe available)
- Care of the infant with a life-limiting condition including early detection and support for respiratory infections
- Feeding – speech and language and dietetic input, nasogastric high-calorie feeds, treat reflux

Notes on spinal muscular atrophy type I

- Autosomal-recessive chromosome 5q12
- Presents in the first year
- On inspection, infant is watchful and alert, but immobile with a frog-like posture, bell-shaped chest with see-saw respirations and proximal limb wasting
- On examination will have hypotonia, muscle weakness, reduced or absent reflexes and fasciculation. Legs more severely affected than arms
- Patients are never able to sit and their lifespan is 12–18 months. Death is usually from respiratory failure
- Diagnosis is essentially a clinical one. Gene probe available

Spinal muscular atrophy type II

Patients with spinal muscular atrophy type II have a later onset of symptoms but most are affected by 12 months of age. They will never be able to stand, but life expectancy is longer – early teens through to middle age depending on respiratory complications. Non-invasive ventilation (eg overnight BiPAP) is required later on in life, and children with this condition will have regular respiratory team reviews.

Physiotherapy, occupational therapy, orthopaedic input and nutritional input (including gastrostomy feeding) also become increasingly important aspects of their management.

Notes on muscle diseases

Myotonic dystrophy (dystrophica myotonica)

Myotonia is the failure of muscle to relax after contraction. Myotonic dystrophy is an autosomal-dominant condition that shows genetic anticipation. The gene has been isolated to chromosome 19 and the involvement of geneticists is essential as the extended family needs advising. The condition is characterised by progressive myotonic weakness, particularly affecting the face, jaws, neck and distal muscles.

The early presentation is as the hypotonic floppy infant (= congenital myotonic dystrophy). The clue is the mother, who is likely to have a relatively expressionless, immobile face and myotonia (failure to relax) when you shake her hand. Older child will present with a myotonic, expressionless, immobile face, hypotonia, poor muscle bulk, weakness, and reduced or normal (but not absent) reflexes. The extent of the motor defect is quite variable.

One important management point is that administration of a general anaesthetic to a patient with myotonic dystrophy (which may be mild and not noted preoperatively) is potentially very dangerous. Gene carriers should have a Medicalert® bracelet for this reason.

Congenital myotonic dystrophy

There is often a history of reduced fetal movements. The infant is floppy from birth – the myotonia presenting later. There is a facial diplegia with a triangular facies. There are often respiratory problems after birth. Associations in infancy include talipes and hip problems. The most severe form presenting in the neonatal period has a mortality of 75% in the first year. In the majority of cases it is the mother who is affected, but it can be the father. Genetic testing is available.

Ataxia

Remember that ataxia can be due to a **cerebellar** or a sensory (**dorsal column**) problem. In a ward or outpatient setting, the most common cause is acute infectious/post-infectious.

Although rare in absolute terms, the commonest cases seen in the examination include:

1. Friedreich's ataxia
2. Ataxia telangiectasia
3. Ataxic cerebral palsy

Differential diagnosis of ataxia

Best split into acute, intermittent and chronic. You are less likely to see an acute case unless there is a shortfall in cases for this station and a patient from the ward is included in the exam.

Acute

- ○ Infectious and post-infectious, eg chickenpox, mycoplasma, measles
- ○ Structural lesions, eg tumours, hydrocephalus
- ○ Drugs, eg phenytoin toxicity
- ○ Metabolic disorders
- ○ Vascular, eg basilar artery thrombosis

Intermittent

- ○ Migraine
- ○ Epilepsy
- ○ Inherited recurrent ataxia (eg Hartnup's disease)

Chronic

- ○ Perinatally acquired (eg neonatal encephalopathy)
- ○ Cerebellar malformation, eg Dandy–Walker syndrome
- ○ Spinocerebellar degeneration, eg Friedreich's ataxia
- ○ DNA repair abnormalities, eg ataxia telangiectasia, xeroderma pigmentosa
- ○ Metabolic, eg Wilson's disease, leukodystrophies, abetalipoproteinaemia, Batten's disease
- ○ Refsum's disease – phytanic acid storage disorder with peripheral neuropathy, deafness and retinitis pigmentosa
- ○ Vitamin E deficiency – abetalipoproteinaemia
- ○ Dysgenesis of the cerebellar vermis, eg Joubert syndrome

Case

Peter is a well-looking 12-year-old boy who on walking has an ataxic gait. He is Romberg's positive. He has pes cavus of his feet, up-going plantar responses and absent ankle jerks.

*The diagnosis is **Friedreich's ataxia.***

I would also like to examine his cardiovascular system.

Notes on Friedreich's ataxia

This is the most common inherited ataxia. It is a progressive disorder with pyramidal tract dysfunction, giving signs of cerebellar degeneration and a peripheral neuropathy. Inheritance is usually autosomal recessive. The gene locus is on chromosome 9. It usually presents before the 15th birthday with increasing ataxia (usual presenting feature) and dysarthria. Because ataxia is sensory, Romberg's sign will be positive. Other features include:

○ Lower limb weakness (pyramidal, ascending)
○ Loss of joint position and vibration sense
○ Absent tendon reflexes
○ Extensor plantars
○ Nystagmus
○ Pes cavus
○ Kyphoscoliosis
○ Cardiac abnormalities (late) include hypertrophic cardiomyopathy, arrhythmias
○ Increased risk of diabetes mellitus – around 10%

Work-up if disease is suspected includes nerve conduction studies, genetic counselling and testing, and ECG.

Treatment is largely supportive. Death is usually secondary to cardiac complications.

Tip +

In an older child with ataxia plus a positive Romberg's sign and up-going plantars, think of Friedreich's ataxia

Ataxia telangiectasia

This condition has autosomal-recessive inheritance; the gene locus is on chromosome 11.

The ataxia is cerebellar and usually presents in early childhood. The telangiectasia is characteristic and usually starts at age 5–6 years. The main areas initially are the bulbar conjunctiva and behind the ears.

A third of these children develop malignancy. There is a 50–100 fold greater chance of developing lymphoreticular malignancy as well as brain tumours.

There is also an increased risk of recurrent infection as there are low levels of IgA and IgG.

Spina bifida

This is a group of spinal developmental disorders in which there is protrusion of the vertebral canal contents through vertebral defects. Spina bifida occurs in around 6 in 10 000 live births. The condition is named after the contents found in the lesion (eg a meningocoele contains meninges and cerebrospinal fluid). Defects can occur anywhere from base of skull to sacrum. The lumbar region is most commonly affected.

Case one

John is a well-looking, 8-year-old boy who is unable to walk alone and uses a wheelchair. He is wearing a nappy. Examination of his lower limbs reveals reduced muscle bulk proximally and distally. There is reduced power and tone in his legs and tendon reflexes are absent. I would like to examine his back, which reveals a healed surgical scar in the lumbosacral area and a scoliosis.

*The diagnosis is **spina bifida.***

Case two

> *Jessica is a well-looking 3-year-old girl who is able to walk with the aid of calipers and a walking frame. She has a waddling gait and neurological signs consistent with a lower motor neurone lesion. Examination of her back reveals a surgical scar over the lumbar region.*
>
> *The diagnosis is spina bifida.*
>
> *I would like to examine her for scoliosis and measure her head circumference.*

Discussion points

- ❍ What associated problems might you expect?
- ❍ How do we attempt to prevent spina bifida?

Notes on spina bifida

- ❍ In all but the mildest sacral lesions there can be some degree of hydrocephalus caused by a hind-brain malformation (Arnold–Chiari type II). This is displacement of the fourth ventricle below the widened foramen magnum, which can cause varying degrees of obstruction of the cerebrospinal fluid as it tries to drain to the basal cisterns. Patients need regular head circumference measurements and need urgent imaging if they present with apnoeas, swallowing difficulties or seizures/impaired conscious level.
- ❍ Folic acid supplementation before conception and during the first trimester has been shown to reduce the risk of spina bifida.

Associated problems

- ❍ Joint contractures, congenital hip dislocation
- ❍ Scoliosis
- ❍ Ulcers on feet (sensory neuropathy)
- ❍ Incontinence of urine and often incomplete bladder emptying (the bladder will be palpable), with a risk of reflux nephropathy. Urology involvement is crucial, especially if there is a poor urinary stream
- ❍ Faecal incontinence with patulous anus, but there may also be neurogenic constipation
- ❍ Learning difficulties in some cases
- ❍ Latex allergy if there are multiple exposures (eg during surgery); around 20% of patients are sensitised

Neurocutaneous syndromes

These are uncommon in clinical practice but are commonly used as short cases in the exam. You need to know the clinical signs in detail and be able to discuss issues around each case:

1. Neurofibromatosis
2. Tuberous sclerosis
3. Sturge Weber syndrome
4. Incontentia pigmenti

Case one – 8-year-old boy

Instructions to candidate

> *Examine this child's skin. What else would you like to examine?*
>
> *Simon has multiple brown macules, which are irregularly shaped. These are café-au-lait spots. He also has axillary freckling. I suspect the underlying diagnosis is **neurofibromatosis type I.***
>
> *I would like to examine his gait and his eyes ...*
>
> *I would like to examine his mother for café-au-lait spots ...*

Discussion points

What other systems might be involved; is there anything else you would like to examine?

Rest of examination in neurofibromatosis

○ Visual fields – examine for evidence of optic nerve gliomata which also cause proptosis and ptosis. Mention that you would refer to an ophthalmologist to examine the eye with a slit lamp for Lisch nodules
○ Blood pressure – measure the blood pressure, palpate the femorals (coarctation of aorta) and listen for renal bruits
○ Head circumference measurement – macrocephaly is common
○ Examine the back for kyphoscoliosis

Notes on neurofibromatosis

Neurofibromatosis is an autosomal-dominant disorder that affects the bone, soft tissues and skin as well as the peripheral and central nervous systems. There are two clearly defined genetic disorders that could be seen in the exam: neurofibromatosis type I (NF-I) and neurofibromatosis type II (NF-II).

Type I (90%)

Autosomal-dominant inheritance, 50% have new mutations. Occurs in 1:2500 to 1:3300 births worldwide.

Gene locus on chromosome 17. Encodes a protein called neurofibromin, which normally acts as a tumour suppressor.

Diagnosed if two or more of the following features are present:

- A first-degree relative with neurofibromatosis
- Axillary or inguinal/perineal freckling (often develops in early puberty)
- Optic gliomas (15%) – higher risk of brain gliomas
- Distinctive osseous lesion, eg kyphoscoliosis, tibial bowing, sphenoid dysplasia
- Two or more neurofibromas or one plexiform neurofibroma. These can be found at any point along a nerve. Can be cutaneous, subcutaneous or plexiform (specific for NF-I). 'Buttonhole' invagination when pressed with a finger is pathognomic
- Two or more Lisch (iris) nodules – hamartomas of iris seen on slit lamp examination
- **Six or more café-au-lait spots greater than 5 mm diameter – Prepubertal child**
- **Six or more café-au-lait spots greater than 15 mm diameter – Postpubertal child**

Tip +

Café-au-lait spots are best defined as brown macules that are irregularly shaped and evenly pigmented. A small proportion of healthy children (less than 1 in 100) have three or more, and one or two may be seen in normal individuals.

The clinical course of NF-I is variable and different systems can be involved as time goes on. Patients with NF-I have an additional 3%–15% increase in their lifetime risk of malignant disease. Optic gliomas, astrocytomas, meningiomas and ependymomas are associated with neurofibromatosis.

Deeper subcutaneous or plexiform tumours need to be monitored as they can undergo malignant change (malignant peripheral nerve sheath tumour) later on in life. This complication rarely presents in the paediatric age group.

Patients are also more at risk of developing phaeochromocytomas, Wilms' tumour and thyroid (medullary) carcinoma.

At least one-third of patients with NF-I will have significant learning disability.

Endocrine problems, specifically short stature and growth hormone deficiency, are more common than in the general population.

Type II (10%)

Autosomal-dominant inheritance, mostly new mutations, gene locus on chromosome 22.

Diagnosis

○ Bilateral acoustic neuromas (also called vestibular schwannoma)

or

First-degree relative with neurofibromatosis type II plus:

○ Unilateral acoustic neuroma or two of the following:
  - Schwannoma (most common tumour in NF-II)
  - Neurofibroma
  - Meningioma
  - Glioma
  - Juvenile posterior subscapular lenticular opacities

Between 60% and 80% of patients suffer from cataracts.

MRI scanning is essential for diagnosis and detection/assessment of CNS tumours.

Management of neurofibromatosis

Management of both types is multidisciplinary; you may be asked to list the health professionals who may be involved in the management of a case:

○ Paediatrician – overall management, assessment at least yearly including checking neurofibromas for signs of malignant change, monitoring growth, plus looking for scoliosis and measuring blood pressure in those with NF-I
○ Paediatric surgeon – excision of neurofibromas that are painful or increasing in size
○ Orthopaedic surgeon – management of kyphoscoliosis
○ ENT surgeon – excision of acoustic neuromas (NF-II)

- Plastic surgeon – correction of facial deformities
- Child psychiatrist/educational psychologist
- Geneticist for counselling as condition has autosomal-dominant inheritance
- Audiologist (NF-II)
- Ophthalmologist (including formal visual field checks, slit lamp)
- Endocrinologist if endocrine tumours in those with NF-I
- Speech and language therapist
- Radiologist to help guide neuroimaging

Remember that this sort of question, whereby you are asked to list the multidisciplinary professionals involved in the care of a complex child, is common.

Case two – 15-year-old boy

Instructions to candidate

Please examine this boy's skin then tell me what else you would like to examine.

> *Christopher has wart-like lesions along his naso-labial folds which I think are adenoma sebaceum. He has several café-au-lait spots on his trunk and a shagreen patch on his lower back.*
>
> *The diagnosis is **tuberous sclerosis.***
>
> *I would also like to use a Wood's lamp to look for depigmented macules, and check his head circumference and blood pressure ...*

Notes on tuberous sclerosis

Autosomal-dominant inheritance with a 50% recurrence risk in offspring. Caused by mutations of the TSC1 and TSC2 tumour-suppressing genes on chromosomes 9 and 16, respectively.

As a result, harmatomas form in multiple organ sites.

Diagnosis is based on major and minor features. Prevalence in children is 1:10 000 to 1:15 000. Up to 70% are new mutations.

Seizures are common (70%), often presenting as infantile spasms. Multiple seizure types have been described.

The overall prevalence of learning difficulties is variable – between 30%–60%. Autism is present in around 25%.

Clinical features/complications

Skin

- Hypopigmented macules – leaf-shaped, best seen with Wood's lamp
- Adenoma sebaceum (facial angiofibromas) are present in 85% of cases over the age of 5 years. Seen as warty lesions along the naso-labial folds. More fully developed when sebaceous glands become hypertrophic at puberty
- Periungual fibromas (present in 20% of cases, and more common in girls)
- Shagreen patches
- Café-au-lait spots

Teeth

- Enamel hypoplasia – 'pits' can sometimes be seen in the enamel

Eyes

- Choroidal hamartoma
- Retinal phakomas

Gastrointestinal

- Rectal polyp

Central nervous system (CNS)

- Giant cell astrocytoma (can lead to hydrocephalus as most often occur in foramen of Monro)
- Tubers
- Subependymal nodules (harmatomas in lateral ventricle wall)

Kidney

- Renal angiomyolipomas
- Polycystic kidneys

Cardiac

- Rhabdomyomas (can cause ventricular outflow tract obstruction and arrhythmias, particularly in postnatal period)

Respiratory

- Interstitial lung disease (lymphangioleiomyomatosis) in some female patients

Investigation

- Regular blood pressure measurements and renal ultrasound, as renal tumours are common

- Echocardiography in neonates/infants and younger children to detect cardiac rhabdomyomas, which then regress in size
- ECG if suspicion of arrhythmias; Wolff–Parkinson–White commoner in this group of patients than in the general population
- EEG useful when there is a suspicion of epilepsy
- Cranial MRI (repeating if later clinical indications or concerns)

Management

Patients should be seen in specialist multidisciplinary clinics as well as at a local level wherever possible. Genetic counselling should be offered and neurodevelopmental follow-up is essential.

Seizures can have variable response to anticonvulsants but rarely with complete seizure control. For infantile spasms associated with hypsarrhythmia on the EEG, Vigabatrin is indicated. Some seizures, particularly as the child grows up, can become refractory to medical treatment; epilepsy surgery can potentially play a role.

Outlook is variable; early death may occur due to seizures or tumours affecting the CNS, heart or kidney.

Patients and families should be signposted to the Tuberous Sclerosis Association, which also has guidelines for clinicians.

Sturge Weber syndrome

In this station you may see a child with a facial naevus. If so, it is important to be able to look for the associated CNS complications seen in some children (around 15%) with a facial naevus. As this is a 9-minute station it is likely that the child will have an associated neurological finding as part of Sturge Weber syndrome. In this disorder there are angiomas (residual embryonal blood vessels) involving the skin of the face and also the leptomeninges.

Notes on Sturge Weber syndrome

Facial naevus (port wine stain)

- Present at birth = capillary haemangioma
- Sporadic
- Unilateral usually, but can be bilateral
- Upper face and eyelid but can be more extensive
- Glaucoma is an occasional complication

CNS associations – not present in all

- Involvement of ipsilateral meninges (leptomeningeal angiomas) and cortex
- Intracranial calcification
- Seizures (in 75%–90% of patients). Results from cortical irritability caused by cerebral angiomata
- Contralateral hemiparesis in around one-third of patients
- Learning difficulties (in up to 90% of patients) and developmental disorders; these are more likely to be present if there is bilateral involvement

Investigation

- Skull X-ray: intracranial calcification – 'rail-track' calcification seen by 2 years
- EEG
- MRI with gadolinium contrast

Management

- Anticonvulsants
- Hemispherectomy/lobectomy occasionally considered as the epilepsy is very difficult to control
- Ophthalmological review and management of glaucoma if present
- Cosmetic – pulsed laser is used for the port wine stain

Case three – 4 year-old girl

Instructions to candidate

Please examine this girl's skin then tell me what else you would like to examine.

> *Kate has hyperpigmented, whorled lesions on her trunk. She has peg-shaped teeth …*
>
> *I think she has incontinentia pigmenti.*
>
> *I would like to measure her head circumference and perform a full neurodevelopmental assessment as well as eye examination, and ask her parents about seizure activity.*

Notes on incontinentia pigmenti/hypomelanosis of Ito

Incontinentia pigmenti

X-linked-dominant disease affecting skin, CNS, hair and teeth. Usually lethal in boys. Clinical features:

- Rash – present in neonatal period as an erythematous bullous rash that fades to pigmented lesions, which are splashed across the skin as if by a paint brush
- Neurological problems (around a third of patients) – seizures/significant learning difficulties/spasticity
- Ocular abnormalities (benign mass in posterior chamber)
- Peg-shaped teeth in some

Hypomelanosis of Ito

This is a distinct syndrome with whorled, marbled areas of *hypo*pigmentation. Half the children have neurological signs and symptoms including seizures, significant learning difficulties and hemiplegia. Most cases are sporadic.

12

MUSCULOSKELETAL AND 'OTHER' STATION

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12.1 MUSCULOSKELETAL

12.1.1 Introduction

Any problem with limbs or movement might be encountered at this station. You will need to be able to examine limbs and joints in general, and the pGALS approach is a key skill which allows you to do this in a systematic way. pGALS refers to paediatric gait, arms, legs, spine. You will also need to be able to examine specific joints in detail, and this chapter suggests an approach for this too. Often, you will be required to adapt your examination to address a specific region or symptom. The Royal College of Paediatrics and Child Health (RCPCH) guidelines suggest that the examiner's directions will be orientated to the type of presenting complaint that would be seen in outpatient departments (eg 'clumsiness', 'difficulty walking', 'pains in legs'). Because the musculoskeletal station is also an 'other' station, you can never be sure that you are encountering a rheumatological problem. If you have a well-practised musculoskeletal examination, then you will be able to adapt it to more obscure symptoms quickly and efficiently. You can then move on to a neurological or developmental examination if you feel that this is more appropriate.

The pGALS screening examination

The RCPCH guidelines suggest that the pGALS (paediatric Gait, Arms, Legs, Spine) approach is the correct way to structure a screening musculoskeletal examination. Full details of this and excellent training materials are available on the Arthritis Research UK website (www.arthritisresearchuk.org). There are video clips of individual examinations (eg of the spine or legs) as well as a complete top-toe examination. Watching these is an extremely valuable use of your precious revision time.

pGALS is a quick and validated musculoskeletal screening tool. It has a high sensitivity for detecting musculoskeletal pathology. All of the joints are examined to screen for pathology. This would normally be followed by a 'look, feel, move' approach if a problem is identified.

Remember that not all musculoskeletal short cases require a pGALS approach. pGALS should be in your repertoire of musculoskeletal examination techniques available for use, but if a clear problem is identified with a joint, you will need to have a plan for how to examine this joint in more detail. The pGALS approach is summarised below.

When to use pGALS in the musculoskeletal station:

1. If general instructions are given (eg 'Examine this child's musculoskeletal system').
2. If you are presented with a history of joint pain, joint stiffness or a limp.

3. If the general instructions ask you to look for the underlying cause of a non-specific symptom (eg 'clumsiness' or 'difficulty climbing stairs/getting dressed/writing'). In these cases the problem could be developmental, neurological or musculoskeletal. Because the pGALS is a quick screening test it is a good place to start if you have limited time available. If no clear pathology is identified with the relevant aspects of the pGALS, you can then proceed to a neurological examination, but you will already have inspected the relevant limbs.
4. You could start with a partial or directed version of the pGALS if you are asked to examine a particular limb or joint. For example, if asked to 'Examine this child's legs', you could start with the Gait section and then move on to the Legs section of the pGALS before becoming more focused.
5. If, during a more targeted short case, you suspect a system condition that affects the joints, you should finish by saying 'I would like to use the pGALS examination to look for any joint pathology elsewhere.' For example, this could be relevant if you suspect inflammatory bowel disease in an abdominal short case or psoriasis in a dermatology short case.
6. If you identify a joint problem or if you are asked to examine one joint and find evidence of any inflammatory arthritic condition, you should ensure that the examiner knows that you would like to look at the other joints. Have in mind which other joints are likely to be affected by the condition that you suspect is present. You may be asked what you are looking for in particular on the pGALS screen.

12.1.2 Clinical examination

The pGALS examination

Top tips

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- ○ Ask the child to copy you performing the manoeuvres (this approach is quicker, clearer to understand and more fun for the child)
- ○ Look for non-verbal signs of discomfort (eg watch the patient's facial expression for evidence of pain, and ask about pain if you notice that a child is hesitant about copying you) and always remember to ask about pain/discomfort before examining a joint
- ○ Look for asymmetry (eg with regard to muscle bulk, range of movement, swelling)
- ○ Comment on any abnormalities that you observe at the time (ie return to a 'look, feel, move' approach after completing the pGALS examination)

Screening questions

- Do you have any pain or stiffness in your joints, muscles or back?
- Do you have any difficulty getting yourself dressed without any help?
- Do you have any problems going up or down stairs?

Standing

- Observe the patient from the front, back and sides:
    - Look at the posture, habitus, rashes, specific features, eg syndrome
    - Look for scoliosis
    - Look at the position of the knees (valgus, varus or neutral)
    - Look for any signs of leg length discrepancy

Walking

- Walking on heels
- Walking on tiptoe
- Look at foot posture; are normal arches present?

Sitting

- 'Hold your hands out straight in front of you':
    - Tests flexion of the shoulders
    - Tests extension of the elbows, wrists and fingers

- 'Turn your hands over and make a fist':
    - Tests supination of the elbow and wrist
    - Tests flexion of the fingers
- 'Pinch your index finger and thumb together':
    - Tests dexterity, co-ordination and function
- 'Touch the tips of your fingers':
    - Tests dexterity, co-ordination and function
- Squeeze the MCP joints:
    - Look for tenderness
- 'Put your hands together palm to palm':
    - Tests extension of the fingers and wrists
    - Tests flexion of the elbow
- 'Put your hands together back to back'
- 'Reach up and touch the sky':
    - Tests shoulder abduction
- 'Look at the ceiling':
    - Tests neck extension
- 'Put your hands behind your neck':
    - Tests shoulder abduction and external rotation
    - Tests flexion of the elbow
- 'Try and touch your shoulder with your ear':
    - Tests lateral flexion of the cervical spine
- 'Put three fingers in your mouth':
    - Tests the temporomandibular joint

Lying

- Feel for knee effusion:
    - Patella tap
    - Cross fluctuation
- 'Bend and straighten your knees' (feel for crepitus):
    - Tests knee flexion and extension
- Hip internal rotation and flexion:
    - Hip flexed to 90°

Standing again

- 'Bend forward and touch your toes':
    - Check for scoliosis
    - Tests forward flexion of thoracolumbar spine

Suggested approach to joint examination

Potential examiner's questions

- This child has joint pains. Examine his musculoskeletal system.
- This child has been complaining of pain in his legs. Examine him.
- This child has difficulty climbing stairs. Examine him.
- These parents have noticed that their child has a limp. Examine her.
- Examine this child's joints.

General approach to joint examination

1. General comments
2. Screening questions
3. Expose the area of interest and comment on any obvious abnormalities
4. Screening passive movements
5. Examine specific joints in detail (use the 'look, feel, move' approach)
6. Look for the impact on function if any pathology is identified
7. Describe the pathology in detail in terms of appearance/range of movement and its impact on function
8. Look for associated pathology in other joints or other systems
9. Be prepared to comment on additional investigations, possible management and the involvement of other health professionals

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Top tips

- Always compare sides
- Don't cause the child pain
- Wherever possible adopt a 'look, feel, move' approach
- If any problem is identified, examine the joint above and the joint below

General observations

○ Comment on the general appearance of the child. Do they look comfortable?
○ Do they look well or unwell?
○ Are there any obvious dysmorphic features?
○ Does the child appear to be well grown?
○ Comment on the presence of mobililty aids (eg wheelchair, orthoses, splint)
○ Comment on any obvious musculoskeletal abnormalities (eg arthrogryposis, hemihypertrophy)
○ Observe the child's posture (are they sitting, standing or lying?)

Screening questions

Always ask about pain and stiffness before you start examining the child. If you cause pain, that may be an instant fail.

Include pGALS screening questions if appropriate.

Symptoms of joint disease

○ Pain
○ Stiffness
○ Swelling
○ Instability
○ Locking
○ Clicking
○ Loss of function

Expose the area of interest

Comment on any obvious abnormalities (eg swollen left knee, swollen interphalangeal joints). If there is an obvious abnormality, this needs to be the focus of the rest of the examination.

Look for muscle wasting, scars, erythema, and asymmetry of appearance or posture.

If you ask the patient to undress, comment on whether they find this easy or difficult.

Undressing patients in the musculoskeletal examination

Always aim to expose fully the area in question. If you think that this will embarrass the patient or cause them discomfort, say to the examiner 'Ideally I would expose the legs/arms/back completely. Are you happy for me to do this?' Because there is a degree of time pressure in the short cases, as a general rule if exposure is relevant to the examination that area of the patient will already be exposed, or dressed in such a way that the area of concern can be quickly and easily exposed.

Screening movements

- Show the child what you would like them to do
- If you have not already done so, now is a good time to introduce the screening parts of the pGALS examination which are relevant to the area that you are examining.

At this stage, if you identify an obvious abnormality using the pGALS movements, this will help to direct the remainder of your examination.

Examine specific joints in detail (use the 'look, feel, move' approach)

Look

- Look for any deformity, erythema, swelling around the joint, scars from previous infection or surgery, and muscle wasting
- Compare both sides
- Comment on posture and/or movements where relevant

Feel

- Watch the patient's face for signs of pain while palpating the joint
- Feel for skin temperature, tenderness and joint effusion

Move

- Ask the child to move the joint before you move it, to ensure that movement is not painful and to get some idea of the range of movement that is possible.
- Compare the affected joint with the normal joint.

Function

- This involves looking for the impact of joint disease, ie how is it affecting the child's day-to-day activities
- For upper limbs, ask the patient to do things which he or she finds difficult (eg drinking from a cup, grasping large objects such as a bottle or box, and small objects such as paper clips). Ask the child to write, draw, comb their hair, take off their shirt, etc
- For lower limbs, observe their gait if you have not already done so
- For the spine, think about dressing and undressing, in particular taking off and putting on shoes and socks

Describe the pathology in detail in terms of appearance and/or range of movement as well as its impact on function.

You will receive marks not only for your examination technique but also for your description of the abnormality that you have found.

> *I examined Claire, who has a swollen left knee. She had a reduced range of movement compared with her right side, and was only able to flex to approximately 100°. There was some crepitus with movement of the knee, and a small joint effusion. There was no obvious muscle wasting. She has a mild leg length discrepancy with a proximal lengthening on the right-hand side. I would like to measure this … She does not have any obvious limp. My impression is that she has a chronic inflammatory arthritis, and I would also like to examine …*

Look for associated pathology

Examine the other joints:

- This is the time to mention the pGALS screening examination if you have not already done so
- If you have found joint pathology, your 'further examination' should include the joint above and the joint below

Systemic disease/extra-articular disease:

- Many rheumatological conditions are associated with physical signs in other areas (eg eyes, skin, cardiac signs). It may be relevant to extend your examination to look for extra-articular manifestations of joint disease. This is covered in more detail in the notes on specific conditions

Remember to look for complications of medical therapy.

Further examination after finding of inflammatory arthritis

- Eyes: blue sclerae, evidence of visual impairment (from uveitis), conjunctival pallor (the patient may be iron deficient as a result of chronic disease or NSAID use, or both)
- Skin: psoriatic lesions, dermatomyositis rash, striae resulting from steroid use, neurocutaneous stigmata, systemic lupus erythematosus (SLE) rash, salmon-pink rash associated with juvenile idiopathic arthritis
- Nails: psoriatic changes, iron-deficiency changes
- Dactylitis: if present this can aid differential diagnosis
- Abdomen: hepatosplenomegaly
- Look for evidence of overgrowth or undergrowth of the affected limb (eg measure for leg length discrepancy)
- Look for evidence of infection if joint pain is known to be of recent onset (consider the possibility of reactive arthritis)

- Check the child's temperature (this is particularly relevant for systemic-onset juvenile idiopathic arthritis)
- Consider growth and nutrition and suggest plotting height and weight if not already mentioned

Further examination after finding of scoliosis

- Skin: look for neurocutaneous stigmata, and scars from previous surgery
- Neurological examination
- Mention pubertal staging: this may be relevant, as scoliosis tends to worsen during puberty
- Respiratory examination if scoliosis is severe (look for evidence of respiratory compromise)
- Look for evidence of Marfan syndrome if the child is tall

Be prepared to comment on additional investigations, possible management and the involvement of other health professionals.

Investigations

- Blood tests – FBC, inflammatory markers, antinuclear antibody, other autoimmune antibodies
- Imaging – consider what imaging would be the most appropriate; plain X-ray, MRI, ultrasound?

Treatment

- Consider both medical and non-medical treatment
- Remember to mention physiotherapy, occupational therapy, orthopaedic involvement and orthotics

Notes on specific joints

Wrists

- Palpate to localise tenderness
- Range of movement: flexion 80°, extension 70°, radial deviation 20°, ulnar deviation 30°

Elbows

- Range of movement: flexion 135°, extension 0°, supination 90°, pronation 90°
- Supination and pronation are tested with the elbows flexed

Shoulders

The range of movement can be easily tested as follows (this method also covers function):

- 'Put your hands above your head':
    - Tests flexion (90°) and abduction (180°)
- 'Give yourself a hug':
    - Tests adduction (45°)
- 'Scratch your back':
    - Tests external rotation (45°)
- 'Hide your hands behind your back':
    - Tests internal rotation (55°) and extension (45°)

Jaw and cervical spine

- Movement at the temporomandibular joint should be examined with the child opening and closing their mouth
- Feel for crepitus over the temporomandibular joint
- Look for acquired micrognathia and dental malocclusion
- Range of movement at cervical spine: flexion 45°, extension 50°, rotation 80°, lateral flexion 40°

Thoracolumbar spine

- Examine the child standing and bending forward for kyphoscoliosis
- Feel for tenderness and check the range of movement: flexion (should be able to touch toes), extension (30° at lumbar area), lateral bending (50° to each side), lateral rotation (30° to each side)
- Functional evaluation: ask the child to pick up an object from the floor or to put on their socks and shoes

Lower limbs

- Examine the gait irrespective of the joint in question (the examination of gait is covered in detail in Chapter 11)
- Remember to look for leg length discrepancy
- Make the child squat (to test for proximal muscle weakness)
- Make the child stand on each leg in turn (to test for Trendelenburg's sign)

Hips

- Note the resting position
- Look for muscle wasting

- Feel for tenderness
- Measure true leg length between the anterior superior iliac spine and the medial malleolus
- Range of movement: flexion 120°, extension 30°, internal rotation 35°, external rotation 45°, abduction 50°, adduction 30°. Stabilise the pelvis before checking the range of movement. Internal and external rotation are measured with the hip flexed, and are the movements most likely to cause pain if there is any hip pathology, so be careful!

Knees

- Look for quadriceps wasting
- Feel for tenderness, raised local temperature and effusion
- Range of movement: flexion 135°, extension up to 10°
- You should know the manoeuvres for checking the stability of the ligaments in the knee. However, they are more relevant to traumatic knee injuries, and you would be very unlucky if you had to demonstrate these in this station

Ankles and feet

- Range of movement at ankle: plantar flexion 50°, dorsiflexion 20°. Inversion (5°) and eversion (5°) occur at the sub-talar joint. Abduction (10°) and adduction (20°) occur at the mid-tarsal joints. The first metatarsophalangeal joint permits plantar flexion (45°) and extension (70°)
- Some joint deformities are more obvious when the patient is weight bearing. When assessing function, ask the child to stand, and observe what happens to their feet and ankles. Looking at their footwear may also provide some information about function. Standing and walking on tiptoe and then on the heels can accentuate problems with the feet

Suggested approach to examining the legs in the musculoskeletal station

'This mother has noticed a problem with her 8-year-old daughter's legs. Examine the child.'

○ General appearance
○ Posture (sitting, standing and lying)
○ Presence of wheelchair, walking aids or orthoses
○ Does the child appear well or unwell?
○ Growth
○ Ask about pain in the legs
○ Expose the legs and look for any obvious abnormality
○ Observe the child's gait (see Chapter 11 [pp.288–290] and pGALS screen [pp.324–325])
○ Consider Gower's sign/squatting to look for myopathy

- If there are no obvious pointers with regard to pathology, perform the 'Legs' section of the pGALS. This will involve asking the girl to lie on the couch, and observing her movements as she does so
- If the pGALS screen is normal, proceed to a neurological examination of the legs
- Look at the spine

Suggested approach to hand examination

This is very important and is the starting point of most systems examinations. You may be asked just to 'look at the hands'. In most cases the features are obvious and lead you on to further examination. In other circumstances a more detailed review of potential pathology in the hand is required, and the following is a suggested scheme.

Instructions to candidate

Examine this child's hands.

Look at this child's hands and describe what you see.

Initial procedure

- Introduce yourself to the child and say 'Please would you roll up your sleeves?'
- Ask whether they have any pain in their hands

Inspection

Look first for any obvious abnormality which, if found, will direct the rest of your examination. Remember to examine both hands at once. If there is no obvious abnormality then work through the sequence below.

1. Inspect dorsum of hands first, looking specifically at:
   - Deformity:
     - Trauma
     - Arachnodactyly – Marfan syndrome
     - Pseudohypoparathyroidism – short 4th, 5th metacarpal
     - Broad hands, stubby fingers, curved little finger, single palmar crease – Down syndrome
     - Large – acromegaly (rare)
   - Asymmetry
   - Muscles and soft tissues:
     - Muscle wasting could point to arthritis, myopathy or neurological disease (for example, combination of claw hand and interosseous wasting points to an

ulnar lesion proximal to the wrist). Remember to look for wasting of the thenar eminence and interosseous muscles in particular

  - Subcutaneous fat and palmar fascia – Dupuytren's contracture (rare in childhood) – indicative of chronic liver disease
  - Blood vessels – Raynaud's phenomenon in primary Raynaud's disease
  - Tendon sheaths – in rheumatoid arthritis thickening of the synovial sheath leads to swelling on the dorsum of the wrist and the front of the fingers and wrist
  - Joints – arthritis can cause anything from mild spindling of proximal interphalangeal joints to gross disorganisation of the hand with subluxation of joints and wasting of small muscles. Psoriatic arthritis involves the distal interphalangeal joints and causes nail pitting.
- Nails:
  - Clubbing – (many causes, you should be very familiar with the list!)
  - Pitting – psoriasis
  - Koilonychia – iron-deficiency anaemia
  - Splinter haemorrhages – (again many causes including bacterial endocarditis, SLE and rheumatoid arthritis)
  - Leukonychia – abnormal zones of nail growth can occur in serious illness, malnutrition and chemotherapy
  - Nail bed telangiectasia or haemorrhages – dermatomyositis, SLE, rheumatoid arthritis, scleroderma
  - Make a point of inspecting the nails from the side to look for clubbing and convex nails
- Skin:
  - Eczema
  - Contact dermatitis
  - Scleroderma
- Posture:
  - 'Claw hand' – hyperextension of the metacarpophalangeal joint and lack of extension of the interphalangeal joint due to weakness of the interossei (damage to the medial cord of the brachial plexus at birth)
  - Hemiplegia – flexed hand, wrist and arm
  - Wrist drop – radial nerve palsy
  - Ulnar deviation of fingers – rheumatoid arthritis
- Stigmata:
  - Neurocutaneous syndromes
  - Evidence of bacterial endocarditis
- Scars – especially preterm venepuncture marks

2. Inspect the palms following same regime as above:
   - Deformity
   - Asymmetry
   - Muscles and soft tissues
   - Skin

- Posture
- Stigmata
- Scars

Palpation

Always ask about pain first:

- Colour, temperature, sweating
- Palpate any swellings to see if they are bony or soft tissues
- You could assess sensation at this point – remember dermatomes and radial/ulnar/median sensory distribution
- Gently squeeze the metacarpal joints to feel for tenderness

Move

1. Comment on tremors/spontaneous movements
2. Range of movement:
    - Metacarpophalangeal joint – flexion 90°, extension 30°
    - Proximal interphalangeal joint – flexion 100°
    - Distal interphalangeal joint – flexion 90°, extension 10°

Tip +

Remember to compare both sides

3. A quick way of assessing hand movements is:
    - *'Make a fist'*: finger flexion at each joint is assessed by looking at symmetry – assesses median and ulnar nerve supply
    - *'Make a star'* (fingers spread out and extended): radial nerve extends and adducts the thumb; the ulnar nerve supplies the intrinsic hand muscles (responsible for abduction) – can test symmetry of finger abduction power
    - *'Make a circle'* (with thumb and index finger): median nerve opposes and flexes thumb – can test symmetry and power of this
    - *'Touch the tips of your fingers with your thumb'*: tests dexterity, co-ordination and function

If there are any abnormalities in the above movements, you can then go on to assess nerves individually in more detail.

Function

The function of the hand is best assessed by simple tests such as:

- Grip strength
- Use of spoon, knife and fork
- Holding a cup

- Drawing or writing with a pencil
- Doing and undoing buttons

Finally

Think about which other areas you might need to examine. There are many systemic conditions with physical signs in the hand, so mention a cardiovascular or abdominal examination if there is clubbing, examination of the skin for a rash if there is any suggestion of psoriasis, etc.

Consider performing a pGALS screening examination if joint pathology is found in the hand.

Mention the need for a neurological examination if any motor or sensory disturbance is found; pay close attention to the brachial plexus and cervical spine.

Nerve injuries with physical signs in the hand

Nerve injuries make good spot cases and should be revised carefully (see Box).

ULNAR NERVE PALSY

- Sensory – loss of sensation of the medial 1½ fingers (ring and little fingers)
- Motor – claw hand, weakness and wasting of small muscles with sparing of the thenar eminence

MEDIAN NERVE PALSY

- Sensory – loss of sensation in the thumb and lateral fingers (up to and including the lateral half of the ring finger)
- Motor – weakness of the thenar eminence, opponens pollicis, lateral two lumbricals, adductor pollicis brevis

RADIAL NERVE PALSY

- Sensory – loss of sensation in anatomical snuffbox
- Motor – loss at the wrist, finger and thumb extension, weakness of extensors of the wrist, thumb, metacarpophalangeal joints of the fingers, 'wrist drop'

Erb's palsy

Caused by injury to C5,6 nerve roots; this most commonly occurs secondary to difficult deliveries, particularly shoulder dystocia. The immediate presentation in the neonate is caused by compression or stretching of the nerves by haemorrhage and oedema, in which case there is a good recovery. Occasionally the nerve is torn (avulsion). Over 90% of cases present at birth will completely recover within the first year of life.

Features:
- Asymmetrical Moro
- Adduction and internal rotation at the shoulder
- Extension at the elbow
- Pronation of the forearm
- Flexion of the wrist (waiter's tip)
- Limb is hypotonic with a reduced biceps jerk

Associations:
- Fractured clavicle

Management:
- Physiotherapy
- Surgery (rarely)

Klumpke's paralysis

Caused by injury to C8 and T1 nerves. It is much less common than Erb's palsy, but also most frequently is a result of childbirth. It classically causes the 'claw hand'. Interossei, thenar and hypothenar muscles are affected. Klumpke's can be associated with Horner syndrome (miosis, ptosis, anhydrosis).

12.1.3 Cases

Case one – juvenile idiopathic arthritis

Instructions to candidate

Please examine Bethany's hands.

> *Bethany is a 10-year-old girl with bilateral swollen wrists. There is painful restricted movement of both wrists. She has marked swelling of the proximal interphalangeal joints of the index and middle fingers bilaterally. Functionally she was unable to unbutton her cardigan. I would like to do a detailed examination of all joints, as well as plot her height and weight on a growth chart.*

This scenario gives you a good opportunity to say what else you would examine.

For example:

- I would like to examine for signs of eye disease
- I would look for evidence of side-effects of treatment (eg cushingoid appearance)
- I would like to examine the skin, looking for any rashes

Case two – psoriatic arthritis; 'hand' examination

Instructions to candidate

Examine John's hands.

> *John is 12 years old. Examination of his hands shows a diffusely swollen right index finger at the distal interphalangeal joint. On closer examination, I can clearly see multiple pits on the nail of this finger. I would like to examine John's other joints. I would also like to examine John's skin, looking for any signs of psoriasis. I would like to plot his weight and height on a centile chart.*

- Joint examination: pGALS screening examination looking for asymmetric oligoarthritis of large and small joints
- Rash: typical of psoriasis. Erythematous papules that coalesce to form plaques with silvery or yellow-white scales. The rash can occur anywhere but is most common on the scalp, knees, elbows and lower back
- Nails may show multiple pits, vertical or horizontal ridging; onycholysis is uncommon
- Eye examination may show chronic uveitis similar to juvenile chronic arthritis

Case three – haemophilia

Instructions to candidate

Examine James' elbow.

> *James is a 10-year-old boy. On inspection, there is no obvious swelling or redness. On palpating the joint, there is no tenderness. On both passive and active movement there is a fixed flexion deformity of the elbow, although the joint is not painful. I note there is also significant bruising to the upper arm.*
>
> *This would be compatible with a diagnosis of* ***haemophilia****.*

Supplementary questions

- ❍ How does arthropathy reflect the seriousness of the haemophilia?
- ❍ How do you treat bleeding into the joint? (Give appropriate infusion of factor, consider immobilisation, ice packs.)

Case four – osteogenesis imperfecta

Instructions to candidate

Examine Clara's eyes please.

> *Clara is an infant, who has blue sclera.*

Supplementary questions

With that in mind, what would you like to examine?

> *I would like to go on and look for bony deformities in the arms and legs, with the diagnosis of osteogenesis imperfecta in mind.*
>
> *The mother may also be affected, so look for any clues.*

This is a very common short case, the main clues being the blue sclera and the presence of the mother in a wheelchair, reflecting the autosomal-dominant inheritance of the condition.

- ○ What different types of osteogenesis imperfecta are there?
- ○ What is the inheritance?
- ○ What is the differential (non-accidental injury)?
- ○ What do you see in the skull of the newborn? (Wormian bones)

Case five – median nerve injury

Instructions to candidate

Examine Steven's hands. He is 10 years old.

> *Steven is a 10-year-old boy who looks well. Looking at his hands, there is atrophy of the thenar muscles and loss of the thenar eminence in the right hand. The hypothenar muscles are well preserved. I would like to do a full motor and neurological examination of his upper limbs ...*

Examine also for:

- ○ Muscle wasting of arms, forearms
- ○ Classic signs of brachial plexus lesions: 'claw hand' in Klumpke's palsy; 'waiter's tip' hand in Erb's palsy
- ○ Classic signs of peripheral nerve lesion: wrist drop in radial nerve palsy; 'monkey hand' in median nerve palsy; and 'claw hand' in ulnar nerve palsy
- ○ Fasciculation indicative of anterior horn cell disease
- ○ Contractures
- ○ Scars in the hand, neck and the back
- ○ Scoliosis

12.1.4 Neonatal hip examination

There is a 'hip' doll available and it is often used in the exam. You should be very familiar with neonatal hip examination from your clinical practice; it is worth quickly revising this topic as it is common to slip into bad practices! Check if your department has one of these mannequins and practise on it. You may be asked about the ongoing management of a baby with a 'clicky' or 'dislocatable' hip.

BARLOW TEST

This is the test for the 'dislocatable' hip (ie femoral head in joint but shallow acetabulum). Stabilise the pelvis with one hand by lying the child on its back with its hips flexed and knees bent. Place the middle finger of your hand over the greater trochanter and the thumb on the inside of the thigh opposite the lesser trochanter. Push down on the hip, creating a posterior force, and you should feel the hip dislocate if abnormal.

ORTOLANI TEST

This is the test to relocate a dislocated hip. The hips are flexed and abducted and the femoral head should move back into the acetabulum.

In practice, the Barlow and Ortolani tests are performed together in one movement, but each side is done separately.

Other physical signs suggestive of developmental dysplasia of the hip are:

- Leg length discrepancy
- Unequal abduction (most valuable)
- Asymmetrical buttock (not thigh) creases (least valuable)

Notes on abnormalities detected on neonatal hip examination

The abnormalities detected fall into different groups:

- Dislocated
- Dislocatable
- Unstable/click

The first two groups require urgent orthopaedic input. Most units will suggest putting the baby in 'double nappies' until they have been seen by an orthopaedic surgeon. Treatment is usually in the form of a Pavlik harness for at least 6 weeks, usually longer.

- It would be sensible to discuss with your local orthopaedic surgeons their strategy for the management of hip problems in the neonatal period. For the baby with a clicky hip, further investigation and follow-up is required.
- In some units ultrasound is offered either to all babies or to babies that have been identified as being at increased risk. Other hospitals do not offer this and there is no consensus as yet.
- The ossific nucleus in the femur appears at 4–8 months; after 8 months, hip dysplasia should be obvious on X-ray.

The risks of a Pavlik harness include:

- Failure (5%): requiring surgical treatment
- Avascular necrosis of femoral head
- Interference with bonding

INCREASED RISK OF DEVELOPMENTAL DYSPLASIA OF THE HIP IS ASSOCIATED WITH

- Girls
- Oligohydramnios
- Breech
- First born
- Torticollis
- Talipes

FAMILY HISTORY IS IMPORTANT

- 1 parent and 1 sibling = 1 in 3 chance
- 1 parent (female child) = 1 in 6 chance
- 1 parent (male child) = 1 in 9 chance
- 1 sibling = next female child has a 1 in 9 chance

12.1.5 Hypermobility

Many people have a few hypermobile joints. Generalised joint hypermobility can be quantitatively assessed using the Beighton score (see Box).

Beighton score

1. Place palms on the ground standing with legs straight (spine hypermobility) – 1 point
2. Extend 5th metacarpophalangeal joint more than 90 degrees (1 point for each side)
3. Oppose thumb to forearm (1 point for each side)
4. Extend elbow more than 10 degrees beyond neutral (1 point for each side)
5. Extend knee more than 10 degrees beyond vertical (1 point for each side)

This gives a total of 9 potential points. A score of more than 4 is suggestive of generalised hypermobility.

Presentation

Hypermobility can present with the following symptoms:

- Arthralgia: most commonly anterior knee pain. Pain is typically felt most when starting or stopping exercise. If knee is involved, then walking downstairs may cause symptoms. Treatment involves strengthening muscles surrounding the joint (swimming is good) and avoiding exercises that cause additional stretch on joint ligaments. Attention should be paid to appropriate footwear and awareness of posture to prevent the development of chronic pain
- Difficulty with gripping – may have difficulty grasping pens/pencils, causing problems with schoolwork/handwriting (wider pencils are easier to grip)
- Delayed walking and history of being clumsy in early childhood

Management is with physiotherapy, occupational therapy and orthotics if required.

Ehlers–Danlos

Group of conditions with different clinical presentation to generalised joint hypermobility primarily because of the following:

- Paper-like scars (tissue fragility with easy bruising): most prominent on forehead and lower limbs
- Skin hyperelasticity (snaps back into place when stretched)
- More extreme joint hypermobility

Ten different types exist – key ones to know are:

- Classical Ehlers–Danlos: severe form, associated with premature rupture of membranes and preterm birth. Patients have a normal life expectancy and may have mitral valve prolapse
- Hypermobility: as the name suggests, the prominent feature is hypermobility and can be difficult to distinguish from generalised joint hypermobility
- Vascular Ehlers–Danlos syndrome: this is rare, features include thin, easily bruised skin, fragile blood vessels that can rupture and cause serious complications and uterine rupture.

Suggested approach to examination of hypermobile joints

Instructions to candidate

This child has joint hypermobility; examine him.

General

- Note body habitus (Marfanoid)
- Look for evidence that hypermobility is affecting function (orthoses)
- Comment on posture (sitting or standing in a way that places strain on joint ligaments)

Hands

- Arachnodactyly
- Ask child to extend 5th metacarpophalangeal joint and oppose thumb to forearm

Arms

- Look for scars/bruises
- Assess brachial pulse character (collapsing)
- Ask patient to extend elbows

Head and neck

- High arched palate?

Chest

- Look for chest wall deformity
- Listen for mitral valve prolapse/aortic regurgitation

Spine

- Can they touch their palms on the floor with straight legs?
- Assess for scoliosis

Legs

- Look for hyperextension
- Look for atrophic scars

Feet

- Look for pes planus (flat feet) – normal up until 6 years, beyond that it may be associated with ligamentous laxity

Gait

- May be abnormal in severe hypermobility

Finally

- Ehlers–Danlos and Marfan syndromes are autosomal-dominant inheritance; you should say you would like to examine parents if you suspect these conditions
- Plot growth if you have not already done this
- Complete cardiac and ophthalmic examination looking for evidence of Marfan syndrome
- Measure arm span, as this is increased in Marfan syndrome

12.1.6 Juvenile idiopathic arthritis (JIA)

This is a very common case. Most patients attend outpatients and physiotherapy only, and are rarely seen on the wards. The use of **methotrexate** and monoclonal antibody therapy has revolutionised the management of chronic arthritis. This means that children seen may have very few physical signs.

These notes are quite thorough; a good knowledge of this potentially confusing area is essential.

Epidemiology

Prevalence is 1:1000 children.

There are a number of different criteria for the classification of juvenile idiopathic arthritis. The following is the **International League of Associations for Rheumatology (ILAR) classification**:

- Age of onset < 16 years
- Disease duration for a minimum of 6 weeks
- Arthritis in one or more joints defined as swelling or effusion, or the presence of two or more of the following signs:
  - limitation of range of motion
  - tenderness or pain on movement
  - increased heat
- *Polyarticular (subdivided into RhF positive or negative)*: five or more joints affected
- *Oligoarticular (subdivided into RhF positive or negative)*: less than five joints affected
- *Systemic disease*: arthritis in one or more joints with intermittent fever of at least 2 weeks duration with at least one of the following:
  - evanescent rash
  - generalised lymphadenopathy
  - hepato/splenomegaly
  - serositis

Polyarticular rheumatoid factor negative juvenile idiopathic arthritis

- Accounts for 25% of juvenile idiopathic arthritis, three times more common in girls; onset can be at any age, but there are two peaks – the first in toddlers (1–2 years of age), and at 10–12 years
- Arthritis in more than four joints in the first 6 months of disease
- Can be symmetrical or asymmetrical, and affects large and small joints
- Temporomandibular joint involvement (even if asymptomatic) can lead to micrognathia or limited mouth opening
- Uveitis is rare, but more likely to occur if ANA is positive
- RF negative, ANA positive in 25% cases
- Prognosis: severe arthritis in 10%–15% cases. Lower likelihood of remission with younger age at diagnosis

Polyarticular rheumatoid factor positive juvenile idiopathic arthritis

- Accounts for 5% of juvenile idiopathic arthritis
- Of patients, 80% are girls; onset usually in late childhood
- Resembles adult rheumatoid arthritis with symmetric polyarthritis affecting upper and lower limbs (typically hands and feet)
- RF positive 100%; ANA 75%
- HLA DR4 suggests more aggressive course
- No uveitis, sacroiliitis rare
- Prognosis: severe erosive arthritis with poor functional outcome in 50% cases. Joint destruction and contractures can occur similar to adult patients unless well treated

Oligoarticular juvenile idiopathic arthritis

- Accounts for 50% of juvenile idiopathic arthritis
- Onset during early childhood, usually under the age of 6 years
- Of patients, 80% are girls
- Tends to affect large joints, most commonly knees and ankles
- Often joints are swollen, warm and tender. They can be stiff and have decreased range of movement
- Can present with swelling but often no pain. The more painful the joint, the more likely an alternative diagnosis
- Hip involvement at presentation is also very rare and should prompt a search for an alternative diagnosis
- Disease-modifying drugs such as methotrexate and infliximab have had a major impact on this condition. This means that children seen in the exam may have very few physical signs

Complications

- **Chronic uveitis** occurs in 30%; often asymptomatic; can cause blindness. Children should be seen regularly by an ophthalmologist for slit lamp examination:
  - ANA positivity (90%) correlates with the risk of uveitis
    Usually other blood tests (including ESR and rheumatoid factor) are normal
  - Active disease is treated with steroid eye drops and mydriatics
  - Chronic uveitis can progress to poor vision with a cloudy cornea and fixed constricted pupil

Prognosis

- Most resolve within 6 months
- Approximately 20% of children who initially do well will have a recurrence (most commonly within the first year)
- Polyarthritis develops eventually in 20% cases. These children are not reclassified as having polyarticular arthritis (although their treatment is

similarly aggressive) if they have four or fewer joints only involved for the first 6 months

Systemic-onset juvenile idiopathic arthritis (formerly Still's disease)

- Accounts for 10% of juvenile idiopathic arthritis
- Usually presents before the age of 5 years
- Affects boys and girls equally
- Systemic features may precede arthritis by months (only 30% of children will have arthritis at presentation)
- Arthritis is usually symmetrical and affects several joints

Differential diagnosis in this condition is important, as systemic JIA is often a diagnosis of exclusion.

Main differential diagnoses are:

- Malignancy (particularly leukaemia)
- Systemic lupus erythematosus
- Kawasaki's disease
- Rheumatic fever
- Infection/sepsis (eg TB, bacterial endocarditis, malaria)
- Reactive arthritis

Clinical features of systemic-onset juvenile idiopathic arthritis

1. Systemic symptoms (can occur before arthritis):

- Fever (> 38.5°C) – needs to be present for at least 6 weeks for definitive diagnosis
- Macular, non-pruritic rash – classically a salmon-pink rash, brought out by fever or heat, often fades as temperature returns to normal

2. Polyarticular arthritis (usually symmetrical)
3. Other features:

- Weight loss/anorexia
- Lymphadenopathy, hepatosplenomegaly
- Pericarditis, pleural effusions
- Anaemia, leukocytosis and thrombocytosis
- Rheumatoid factor (RF) and antinuclear antibody (ANA) negative
- No uveitis or sacroiliitis
- Myalgia/arthralgia

Enthesitis-related arthritis

- Accounts for 10%–15% of juvenile idiopathic arthritis
- Of patients, 90% are boys
- Onset during late childhood
- Enthesitis refers to inflammation of tendon insertions; typically the Achilles tendon and plantar fascia are involved
- A few large joints are involved, especially the hip girdle; sacroiliitis is common
- Many develop ankylosing spondylitis
- Associated with inflammatory bowel disease (ask about family history)
- Sometimes see 'sausage finger' (dactylitis)
- Acute uveitis is seen in 10%–20% cases. ANA and RF negative
- HLA B27 positive in 75% cases

Juvenile idiopathic arthritis – management

How to manage juvenile idiopathic arthritis is a question that is commonly asked. This is a good opportunity to show the examiner that you appreciate the multidisciplinary management of a complex and chronic condition such as juvenile idiopathic arthritis.

Management of juvenile idiopathic arthritis

DRUGS

- Non-steroidal anti-inflammatory drugs remain the first line, particularly ibuprofen. Doses are usually high and antacids are occasionally required for gastrointestinal side-effects
- Corticosteroid injection – useful in oligoarticular disease. They are increasingly used early in severely inflamed joints with good results
- Systemic corticosteroids in severe disease – particularly disease with systemic manifestations
- Disease-modifying drugs such as methotrexate (other less-used options include ciclosporin)
- Anti-TNF monoclonal antibody therapy is widely used

NON-DRUG TREATMENT

Physiotherapy and occupational therapy

- Hydrotherapy
- Passive movement
- Gentle exercises
- Splints

12.1.7 Other conditions

Psoriatic arthritis

Epidemiology

- Chronic inflammatory arthritis before 16 years of age, preceded by, accompanied by or followed within 15 years by psoriasis
- Prevalence is expected to be 10–15 cases per 100 000
- Age at onset usually around 6 years
- F > M = 2:1

Diagnosis

- The diagnosis is clinical, according to Vancouver criteria (see Box)
- Psoriatic arthritis should be suspected in chronic arthritis with dactylitis or asymmetric involvement of large or small joints, particularly the first metatarsophalangeal or the distal interphalangeal joints
- Rheumatoid factor is negative
- ANA may be seen in 17%–50% cases (there is also a risk of uveitis)

Vancouver criteria for diagnosis

DEFINITE PSORIATIC ARTHRITIS

- Arthritis with typical psoriatic rash
- Arthritis with three of the four following minor criteria:
  - Dactylitis
  - Nail pitting or onycholysis
  - Psoriasis-like rash
  - Family history (first- or second-degree relative) of psoriasis

PROBABLE PSORIATIC ARTHRITIS

- Arthritis with two of the four minor criteria

Management and prognosis

- Management is along the same lines as for juvenile idiopathic arthritis
- Many children with pauciarticular disease may progress to polyarticular disease, some requiring major reconstructive surgery of hips and other joints
- Course may be relapsing and remitting with or without flaring of skin disease

Juvenile dermatomyositis

This is an unusual case and a rare condition in clinical practice, but has come up in the exam:

- The main symptoms are muscle weakness, lethargy and a rash
- Painful tender muscles and proximal muscle weakness usually involving the shoulder girdle or lower limbs
- Rash – faint purple rash over the upper eyelids and cheeks (butterfly), a different type of rash can affect the knuckles (Gottron's papules), elbows and knees, which is more erythematous dry and scaly
- Can get dysphagia (adults), skin calcinosis
- Raised ESR , CPK and LDH
- MRI will detect muscle inflammation
- Diagnosis is confirmed by muscle biopsy
- Treatment is with steroids ± other disease-modifying anti-rheumatic drugs (such as methotrexate, azathioprine, cyclophosphamide)

Osteogenesis imperfecta

Osteogenesis imperfecta is a disorder of connective tissue characterised by bone fragility. The disease encompasses a phenotypically and genetically heterogeneous group of inherited disorders that result from mutations in the genes that encode for type 1 collagen. There are at least eight different forms (type I to VIII). Its manifestations vary from perinatal lethal forms, to moderate forms with deformity and a propensity to fracture, to clinically silent forms with subtle osteopenia and no deformity. Type I is the mildest form and type II the most severe.

Most cases are sporadic mutations or autosomal dominant and so it is important to take a detailed family history.

Medical treatment is with cyclical pamidronate. Aggressive orthopaedic treatment is the mainstay of treatment, aimed at prompt splinting of fractures and correction of deformities. Genetic counselling may be important. Reliable prenatal diagnosis is not available for all forms of osteogenesis imperfecta, although severely affected fetuses may be confidently recognised by X-rays and ultrasound screening.

Hemihypertrophy

This is often difficult to recognise. It may involve the whole of one side of the body, or be limited in extent, eg to just one leg. It may be congenital, in which case the tissues are structurally and functionally normal. It may be an isolated finding, but it has been associated with learning difficulties, ipsilateral-paired internal organs and rarely, with Wilms' tumours or adrenal carcinomas. Hemihypertrophy can be confused with regional overgrowth secondary to neurofibromatosis type I, haemangiomas and lymphangiomas. Hemihypertrophy occurs in 25% of patients with Beckwith–Wiedemann syndrome.

There is an increased risk of tumours, and so you may be asked about further investigations. You should mention regular abdominal ultrasound scanning to screen for Wilms' tumour. You could also mention genetic testing and orthopaedic referral.

Miscellaneous conditions

Short neck

There are different causes of a short neck, and you may come across a child in the exam with a short neck and need to be aware of the differential diagnosis:

- Turner syndrome
- Skeletal dysplasia
- Klippel–Feil syndrome

Plagiocephaly

This is an increasingly common referral for paediatric input, almost certainly because of the 'Back to Sleep' campaign. Babies are put to bed lying on their backs, and so their skulls can become moulded if they always sleep in the same position. Most plagiocephaly is simple ('positional'), and improves as the child develops and isn't lying down as much; increased coverage with hair also helps the appearance. The use of 'helmets' to mould the skull is controversial and the general opinion is that these are not recommended.

It is essential however to be aware of some of the more common craniosynostoses and the relevant syndromes, including Crouzon and Apert.

Bow legs

This is often a normal finding until the age of 18 months. Think about symptoms and signs of **rickets** if it is extreme or persistent.

SIGNS OF RICKETS

- Delayed dentition, dental caries
- Delayed closure of the anterior fontanelle
- Bow legs
- Swelling of epiphyses at the wrists and ankles
- Curving of the long bones
- Craniotabes
- Kyphoscoliosis
- Pathological fractures
- Rickety rosary
- Myopathy

INVESTIGATIONS FOR RICKETS

Calcium levels, phosphate levels, urinary calcium:creatinine ratio, alkaline phosphatase (ALP) levels, parathyroid hormone levels, vitamin D levels, plain X-ray of affected joints

Toe walking

- Can be a normal finding up until age 3
- Neurological disorders include cerebral palsy, Duchenne muscular dystrophy, spinal cord problems, congenital tendo-achilles shortening
- Think about possible leg length discrepancy if unilateral
- Habit

Flat feet

This is a normal finding in most young children. It is associated with flexibility of the forefoot, and normally resolves by the age of 6 years.

Intoeing

This can be due to:

- Femoral anteversion (patella faces inward, causing a 'knock-kneed' appearance)
- Tibial torsion (patella faces forward)
- Metatarsus adductus ('C'-shaped curve to the lateral aspect of the foot).

If the child is neurologically and developmentally normal, the parents can be reassured that the condition will improve with time. Encouraging active play can strengthen the leg muscles and reduce the severity. The child should be discouraged from sitting with their legs in a 'W' position.

Foot drop

- Variety of causes
- Patient has a stepping gait and lifts the affected limb high to avoid scraping the foot on the floor. They are unable to walk on their heel
- Possible causes:
    - Lateral popliteal nerve palsy (look for signs of injury below and lateral to the affected knee)
    - Peroneal muscle atrophy (look for hereditary motor and sensory neuropathy/Charcot–Marie–Tooth's disease)
    - Poliomyelitis

Talipes

- Positional talipes occurs due to the in-utero position, and requires, at most, some parental physiotherapy
- Non-positional talipes or clubfoot is a multifactorial deformity of primarily genetic origin; 30%–50% require casting only. The cast is applied within the first week and then changed weekly. Surgery is reserved for those not responding to casting. If surgery is required, usually at 6–9 months, it involves lengthening the tight tendons and releasing tight joints with pinning and casting – usually with good results. There is a 3% recurrence risk in future children.

Orthoses

Children with neurological disorders are very prone to developing joint contractures and bony abnormalities, particularly spinal kyphoscoliosis. Orthoses are useful in helping to prevent contractures, in maintaining a good position if contractures have been repaired and in providing joint stability. They are particularly useful in aiding individual children with mobility. The type of orthosis depends on the child's individual needs, eg they may be ankle–foot orthoses if there is just ankle and foot involvement, extending to the knee if the knee is involved. Should there be a scoliosis, thoracolumbar orthoses are available. Wrist orthoses are also commonly used.

Perthes' disease

- Arises from disturbance of the upper femoral epiphysis secondary to ischaemic necrosis of unknown aetiology
- Onset commonly between 4 and 9 years. Bilateral in 15% cases
- Earliest sign is a limp, usually accompanied by hip or groin pain. Initial symptoms may be intermittent, causing diagnostic confusion, especially if the initial X-rays are normal
- X-rays should be AP and frog-leg lateral. Initially the ossified portion of the femoral head appears smaller compared to the normal femoral head. Following this, the ossific nucleus becomes more radio-opaque, then the

epiphysis develops a fragmented appearance. This is followed by a gradual return to normal with new bone formation

- Differential diagnosis is septic arthritis, osteomyelitis, transient synovitis, post-traumatic fracture, neoplasia, slipped upper femoral capital epiphysis
- The short-term aim of treatment is to reduce pain and hip stiffness
- The disease is a self-limiting process and may last from 2 to 4 years
- Restriction of activity helps to relieve the pain; occasionally pain may be so severe as to warrant a brief period of non-weight bearing with crutches and bed rest
- The long-term goal is to reduce residual deformity. Controversy exists as to the best way of doing this. The important principle is femoral head containment so that re-ossification produces a spherical femoral head. This can be achieved by non-operative bracing in abduction, or by operative means
- Treatment method depends on the age of the child, severity of involvement of the femoral head and the expertise of the local orthopaedic surgeon
- Prognosis is best for young children (under the age of 5), with full recovery and no disability – the remainder are left with a degree of residual disability of varying severity.

12.2 Haematology

12.2.1 Introduction

Many haematological conditions are encountered during the examination of other systems, for example:

- Skin: purpura in Henoch–Schönlein purpura
- Joints: swollen joint in haemophilia
- Abdomen: splenomegaly in hereditary spherocytosis
- General appearance: short stature, malar hypertrophy in thalassaemia

It may be that you will be asked to examine the *haematological system*, and if you are, then remember that the examiner is looking for a clinical assessment of the following:

- Dysmorphic features on inspection which point to a diagnosis with haematological implications
- Signs of anaemia, extramedullary haematopoiesis (bones, liver, spleen, rarely skin) that would indicate red cell disease
- Platelet or clotting factor deficiencies and their complications
- Complications from underlying disease or treatment, such as sequelae of immunodeficiency

You will not only be looking for a diagnosis but also seeking some evidence of:

- Severity
- Whether or not any complications have already occurred (eg cholecystectomy for spherocytosis, neurological complications in sickle cell disease)

12.2.2 Terminology

The **reticuloendothelial** system is the mononuclear phagocyte system and consists of cells in the bone marrow, peripheral blood and tissues highly specialised for the function of endocytosis and intracellular digestion. It plays a role in the inflammatory response and acts as the main line of defence against bacteria in the bloodstream, controlling the haematogenous dissemination of organisms.

Hypersplenism is a clinical syndrome in which normal splenic function becomes excessive as the spleen and its mononuclear phagocyte tissue enlarge. Pancytopenia usually occurs.

The most common cause is venous obstruction, such as that seen in extra-hepatic venous obstruction.

Hyposplenism refers to diminished splenic function associated either with the absence of the spleen (congenital or following splenectomy) or functional impairment, eg sickle cell disease. The main consequence of hyposplenism is the risk of overwhelming bacterial septicaemia. Prophylactic penicillin and pneumococcal and meningococcal immunisation are required.

12.2.3 Clinical examination

Suggested approach to haematological examination

Initial procedure

Introduce yourself.

General

- Comment on growth and whether the child looks ill or well
- Comment on ethnicity (particularly if thinking of thalassaemia, hereditary spherocytosis or sickle cell disease), as this is highly relevant; comment on the parents' ethnicity as well if you think it is appropriate
- Comment on obvious pallor or jaundice; remember that haemochromatosis will lead to 'bronzing'
- Note dysmorphic features, and inspect upper limbs, as TAR syndrome (thrombocytopenia, absent radius) and Fanconi syndrome are associated with upper limb abnormalities
- There may be obvious bruising, petechiae or purpura

Hands and arms

- Clubbing
- Koilonychia
- Scars from cannulae
- Pallor
- Assess pulse rate and volume: if there are any concerns, then check blood pressure (BP), otherwise you could mention checking BP later (think symptomatic anaemia; although this would be uncommon in the exam scenario, you may be asked to talk about it)

Head and neck

- Frontal bossing, malar prominence
- Eyes (pallor or jaundice, conjunctival haemorrhage)
- Lymphadenopathy
- Lips (angular cheilitis)
- Look at gums (hypertrophy in neutropenia) and look at the dentition
- Tongue
- Tonsils

Chest

- Hickman line or Port-a-Cath®
- Heart murmur (flow murmur from anaemia)
- Comment on the presence or absence of increased work of breathing (think sickle chest syndrome, which is unlikely to present to clinical exams but is an important part of the examination)

Abdomen

- Scars, including laparoscopic (think splenectomy, cholecystectomy)
- Bruises from injection sites
- Liver and spleen
- Inguinal lymph nodes

Legs

- Signs of infection
- Scarring or bruising
- Joint disease (think haemophilia)

Finally

- Axillary lymph nodes – very difficult if children are even slightly ticklish, so leave until the end, but don't forget about it
- In a boy you would normally palpate the testes; indicate to the examiner that you would complete the examination by doing this (looking for testicular lymphoma or testicular relapse of haematological malignancy)
- Neurology – especially for sickle cell disease
- Check temperature (crucial for neutropenic or immunodeficient patients)
- Mention that you would like to plot height and weight on a growth chart
- Do parents have evidence of disease?

12.2.4 Cases and conditions: miscellaneous

Lymphadenopathy

It is important to have a differential diagnosis if you find lymphadenopathy. This will depend upon whether the lymphadenopathy is localised or generalised. Cervical nodes are, for example, very common in children with either current or recurrent upper respiratory tract pathology or facial/neck eczema.

In a child with a large, unilateral axillary node, ask about encounters with cats/kittens (think cat scratch disease).

Tip +

Normal lymph nodes do not exceed 2.5 cm in diameter

Simple differential diagnosis of generalised lymphadenopathy

- Infection – infectious mononucleosis, cytomegalovirus, HIV
- Autoimmune disorders
- Leukaemia/lymphoma
- Storage diseases (rare)

Investigation of lymphadenopathy

There are NICE guidelines for when it is appropriate for GPs to refer children with lymphadenopathy for a paediatric opinion (when there is no evidence of local disease) at www.nice.org.uk. Look up Clinical Guideline CG27 (*Referral Guidelines for Suspected Cancer)* which gives the following indications for further investigation:

- Non-tender, firm or hard lymph nodes
- Lymph nodes > 2 cm
- Lymph nodes progressively enlarging
- Other features of general ill health, fever or weight loss
- Axillary node involvement (without local disease such as dermatitis or infection)
- Supraclavicular node involvement

Investigation of lymphadenopathy largely depends on the history and physical characteristics of the enlarged node. History taking and physical examination should also focus on excluding serious causes of lymphadenopathy.

Investigations may include:

- 'Watch-and-wait' with regular review and safety-netting
- Full blood count (FBC) and film, monospot if acute lymphadenopathy
- Lactate dehydrogenase (LDH), erythrocyte sedimentation rate (ESR)
- EBV serology, Bartonella serology (cat scratch disease), if axillary node
- Imaging – looking either at the node itself or for evidence of lymphoma elsewhere (chest X-ray, abdominal CT/ultrasound)
- Biopsy

Purpura

The finding of purpura reflects a bleeding tendency.

If purpura are seen in the exam it is important to remember the differential diagnosis:

- Infection – meningococcal disease
- Thrombocytopenia – neonatal thrombocytopenia, idiopathic thrombocytopenic purpura (ITP), leukaemia
- Coagulation disturbance – disseminated intravascular coagulation, haemophilia, von Willebrand's disease
- Vasculitis – Henoch–Schönlein purpura (HSP)

Children with purpura are unlikely to be acutely unwell with meningococcal disease, but may be shown in the recovery phase if there is a shortfall of patients. All children with purpura need a full systemic examination with particular focus on the reticuloendothelial system to elicit a cause. In addition, it is sensible to mention basic investigation, which includes a FBC, blood film and clotting screen.

Henoch–Schönlein purpura

Instructions to candidate

Look at this child's legs; what do you see?

Case

> *Jenny looks well. On inspection of her legs I can see a purpuric rash, limited to the extensor surfaces and buttocks. On palpation it does not blanch with pressure. She also appears to have a swollen left ankle, not hot to touch, not red, not tender, but with limitation of movement in all directions. The right foot does not appear abnormal and … abdominal palpation reveals no masses and no tenderness*
>
> *… I would like to take her blood pressure and a urine sample …*

What is your diagnosis?

Henoch–Schönlein purpura.

What is the differential diagnosis?

- Other vasculitis, eg systemic lupus erythematosus
- Thrombocytopenia, although this wouldn't explain the swollen joint

- Sepsis, but the child does not look unwell
- Could be leukaemia

List clinical features/problems

- Purpuric rash (urticaria can also occur), mostly on extensor surfaces anc buttocks, in the absence of thrombocytopenia or coagulopathy
- Joint involvement: arthritis or arthralgia (mainly affecting the lower limbs) ir 60%–80% of cases
- Gastrointestinal involvement: including colicky abdominal pain, vomiting, gastrointestinal bleeding, and rarely intussusception, pancreatitis
- Renal involvement in around 40%, including microscopic haematuria, frank haematuria and proteinuria (check the urine protein to creatinine ratio), nephrotic/nephritic picture, renal failure and hypertension
- Neurological involvement is recognised, but usually mild
- Lung involvement is rare
- Testicular involvement may manifest as uni/bilateral orchitis

Notes on specific conditions

Henoch–Schönlein purpura (HSP)

Commonest systemic vasculitis of childhood with incidence of up to 20:100 000 children per year. Half of cases occur in children under 5 years, 75% in those under 10 years. Commoner in boys and in winter months. Immune mediated; often there is a history of preceding viral upper respiratory tract illness. There are no specific laboratory diagnostic tests. Skin biopsy shows typical features but is not routinely performed in the UK. Urine should be dipped for haematuria and/or proteinuria, and the serum creatinine concentration should be noted. If there is persistent proteinuria with a markedly elevated urinary protein to creatinine ratio, the child should be discussed with a paediatric nephrologist, according to local protocols.

Treatment remains controversial. Use of steroids for abdominal pain is generally well accepted, although there are concerns that steroids may mask a worsening gastrointestinal picture or a lymphoproliferative disorder. Joint symptoms should be treated as required with analgesia and rest. The treatment of renal complications may require the help of a paediatric nephrologist. All patients with significant renal involvement require long-term follow-up. Long-term morbidity is mainly due to renal disease.

Relapses of HSP can occur.

Idiopathic thrombocytopenic purpura (ITP)

This is an autoimmune condition in which circulating platelets are destroyed; it is often triggered by a viral infection. It is relatively rare, with an incidence of around 4:100 000 children. It usually presents with multiple petechiae and purpurae as the platelet count falls. Course is usually self-limiting and benign. If it lasts over 6 months it is termed chronic ITP. Most cases can be diagnosed by excluding other causes of thrombocytopenia (clinical history, examination, FBC and film). Platelet count is typically very low.

More significant bleeds such as menorrhagia, gastrointestinal haemorrhage and epistaxis are rare, with a prevalence of no more than 5%. Of greatest concern to patients and parents is the risk of intracranial haemorrhage, which is very rare, occurring in 0.2%–0.8% of patients in large case series. Oral steroids or intravenous immunoglobulin are occasionally used in severe disease. Teenage girls with menorrhagia will require treatment with antifibrinolytic agents.

Some patients with chronic ITP will need to undergo a splenectomy, but many experience remission without this. Monoclonal antibody therapy (rituximab) has shown promise in refractory cases.

Other causes of thrombocytopenia that may be seen in exam are shown in the Box.

Wiskott–Aldrich syndrome

- X-linked condition
- Severe eczema, thrombocytopenia, immunodeficiency

Thrombocytopenia, absent radius (TAR) syndrome

- Autosomal recessive
- Absent radii is the most common finding
- Cardiac, renal or other skeletal malformations may occur

Anaemia

You may be shown a child who is obviously pale and be asked to show the examiner features of anaemia (see Box), and to come up with a differential diagnosis. The cause may range from iron deficiency (common) to red cell aplasia (very rare), but marks will be gained for conducting a focused examination and giving a sensible differential.

Clinical features of anaemia

○ Patient may look pale and tired – comment on this
○ Is patient jaundiced? Will narrow down differential diagnosis
○ Hands – koilonychia (spoon-shaped nails)
○ Mucous membranes (look at conjunctiva) and nail beds for pallor
○ Flow murmur

Tailor the rest of your examination to initial findings, eg if jaundiced, look for other signs of haemolysis and examine for spleen, etc.

Notes on anaemia

Anaemia is a reduced (2 SD below mean) haemoglobin level for the age and sex of that individual. In the under 5s it has been defined by the WHO as an Hb of < 11.2 g/dl.

It will be broadly due to defective production, loss or excessive destruction of red cells (see below).

Causes of anaemia

Defective production/maturation of red cells:
○ Iron deficiency
○ Chronic disease with decreased erythropoietin production, prematurity
○ Vitamin B_{12} and folate deficiency
○ Marrow failure – aplastic anaemia, infiltration, pure red cell aplasia (Diamond–Blackfan), drugs

Loss of red cells:

- ❍ Haemorrhage, causes of which may include:
    - menstruation
    - non-steroidal anti-inflammatory drugs (NSAIDs)
    - inflammatory bowel disease
    - peptic ulceration
    - non-IgE mediated cow's milk protein allergy with colitis
    - Meckel's diverticulum
    - hookworm infestation (a very common cause of blood loss in some parts of the world)
- ❍ Excessive destruction:
    - Haemolytic anaemias

Initial investigations to aid in narrowing down the differential diagnosis are blood film (for red cell morphology), reticulocyte count (increased if loss or destruction, decreased if defective production) and iron indices, in particular ferritin. Haemoglobin electrophoresis is warranted if a haemoglobinopathy is suspected.

Iron-deficiency anaemia

This is the commonest cause of anaemia in children, and is present in around 15%–40% of inner-city preschool children in the UK. You may get a case in the exam if it is severe enough to show signs. Occurs when there is insufficient iron for erythropoiesis, especially in the context of increased demand during the rapid growth phase from 6 months to 3 years combined with a suboptimal diet. For this reason there is also a peak in incidence during the adolescent growth spurt.

Clinical problems arising from iron-deficiency anaemia are many and include:

- ❍ Faltering growth
- ❍ Decreased exercise tolerance/stamina – 'tired all the time'
- ❍ Decreased attention span and so learning ability
- ❍ Pica
- ❍ High-output cardiac failure if very severe

You may be asked how to address iron deficiency in infants. Current advice is to breastfeed (or use an iron-fortified formula) then wean onto iron-containing foods by 6 months of age. Cows' milk 'off the shelf' should be avoided in infants for many reasons including the low iron content; monitoring from a paediatric dietician (if available) may be required. Appetite improves as the condition is treated.

Fanconi anaemia

Very rare in actuality but if there is a patient in the region, they may well be invited to the examination – always remember that you can see rare conditions in the examination.This is an autosomal-recessive aplastic anaemia with a pre-malignant potential. There are characteristic physical findings in 50%–75%. Bone marrow failure with resultant pancytopenia develops towards the end of the first decade.

Characteristics

- Short stature
- Hyperpigmentation +/– café-au-lait spots
- Microcephaly
- Microphthalmia
- Triangular facies
- Hypoplasia/aplasia of the thumb
- Renal anomalies
- Skeletal anomalies
- Learning difficulties

Hereditary spherocytosis

This is a very common clinical case in the examination.

> ***Tip***
>
> Northern European child plus pallor, jaundice and splenomegaly = hereditary spherocytosis until proved otherwise

Instructions to candidate

Examine this child's abdomen; why is he jaundiced?

This boy presents with abdominal pain, pallor and a palpable spleen. What are your thoughts? What would you do?

Clinical findings

○ Northern European child
○ Looks well
○ Jaundice (mild)
○ Pallor
○ Cholecystectomy scar (occasionally)
○ Splenomegaly or a splenectomy scar (scars may be laparoscopic)

Case one

> This is a well-looking boy. I would like to examine him. His conjunctivae are pale and he is mildly jaundiced. He has a palpable spleen. There are no other abnormalities. I suspect that he has *hereditary spherocytosis* …

Case two

> This is well-looking boy. There is no pallor or lymphadenopathy. He has a cholecystectomy and splenectomy scar. The underlying diagnosis is most likely to be *hereditary spherocytosis* …

Notes on hereditary spherocytosis

Autosomal-dominant condition. Incidence is 1 in 5000 in Northern Europeans. Usually presents in childhood with the classical triad of anaemia, jaundice and splenomegaly. The jaundice is unconjugated and worsens during infections. Pigmented gallstones are present in 85% by the second decade. Aplastic crises can occur.

There are guidelines on the diagnosis and management from the British Committee for Standards in Haematology (www.bcshguidelines.com). The diagnosis is based on the combination of a typical clinical history (+ family history), examination, and FBC and blood film. In a typical case there will be spherocytes seen on the film and an increased number of reticulocytes.

Treatment is with folic acid supplements (unless mild) with regular monitoring of clinical state and blood picture.

In severe cases, particularly where symptoms (eg from gallstones, significant anaemia) are impacting on quality of life, **splenectomy** is considered. The spleen is the site of red cell destruction and its removal will reduce haemolysis and reduce the incidence of gallstones. Cholecystectomy is occasionally required, particularly if there are already symptomatic gallstones.

Once a child is asplenic there is an increased risk of pneumococcal and other infections. Pneumococcal, meningococcal and haemophilus influenza type B immunisation, and lifelong prophylactic penicillin need to be given.

Differential diagnosis

Other causes of haemolytic anaemia include:

- Hereditary elliptocytosis
- Pyruvate kinase deficiency
- Autoimmune haemolytic anaemia

Thalassaemia

Thalassaemia and the different variants are often seen in the exam. If you work in a hospital where such cases are rare, then it is essential to read about and try to get to see some cases.

Case

> *This is a well-looking adolescent boy of Asian origin who looks well grown. On inspection of his hands, he has multiple scars from iv cannulae. His sclerae show a tinge of jaundice and on examination of his abdomen, he has a palpable spleen … I also noticed that he had some patches of depigmentation on his abdominal wall.*
>
> *I think this child has* ***thalassaemia major*** *… I would like to plot his height and weight on a growth chart and check his pubertal status.*

Clinical findings

- ○ Mediterranean/African/Indian/SE Asian child
- ○ Multiple iv cannula sites in the hands, may be a Port-A-Cath®
- ○ Jaundice (mild), pallor
- ○ Multiple depigmented patches on abdominal wall from subcutaneous desferrioxamine administration
- ○ Palpable spleen

The above refers to a well-controlled child with thalassaemia on a regular transfusion regime. Children who have been severely affected may have signs of *extramedullary haemopoiesis* (see notes below):

- ○ Frontal bossing
- ○ Malar hyperplasia
- ○ Hepatomegaly
- ○ Splenomegaly
- ○ Previous long bone fracture sites

Notes on thalassaemia

The thalassaemias are a heterogeneous group of recessively inherited disorders characterised by deficient synthesis of either alpha or beta chains of haemoglobin. There are various different types and there are approximately 100 different mutations known to cause thalassaemia, many of which are unique to localised geographical areas.

These notes are on the **transfusion-dependent (major) form of beta-thalassaemia.** This occurs in the populations mentioned above and is often diagnosed antenatally or after routine newborn screening tests (Guthrie).

There is a globin chain imbalance that results in ineffective erythropoiesis and a severe progressive haemolytic anaemia during the second 6 months of life. The anaemia is microcytic and hypochromic, with poikilocytes and target cells on the peripheral blood film. There are large numbers of nucleated red cells and a marked increase in HbF. There is iron overload, with increased absorption of iron from the GI tract.

Clinical features vary with severity.

Undiagnosed children who have become severely affected can present with pallor, jaundice, faltering growth, feeding difficulties and hepatosplenomegaly.

In untreated or inadequately transfused patients, there is proliferation of medullary and extramedullary haemopoiesis with massive expansion of the skull and face causing characteristic facies, frontal bossing is a classical sign of this.

Bones can become affected by osteopenia and osteoporosis, and children can have pathological fractures.

Longstanding haemolysis can give rise to gallstones.

Iron overload (which can occur before, or as a consequence of transfusion) is a particular problem in children with thalassaemia, giving rise to the following:

- Endocrine problems. These become commoner after multiple transfusions. Growth can be impaired and puberty delayed. Hypothyroidism and hypoparathyroidism can occur. Diabetes mellitus can occur due to pancreatic siderosis
- Iron deposition in the liver can cause cirrhosis
- Cardiac failure can occur due to cardiac siderosis

With the modern technique of management many of these complications can be avoided, and others ameliorated and their onset delayed.

Treatment

Aims to control the ineffective erythropoiesis and so reduce the likelihood of hypersplenism and its complications. Guidelines on the management of thalassaemia can be found on the website of the UK Thalassaemia Society (www.ukts.org):

- **Hyper-transfusion regime:** blood transfusions on a regular basis (eg monthly) to maintain haemoglobin above 10 g/dl. Decision to start is based on regular assessment of clinical problems (particularly emerging features of bone/cardiac involvement) and growth.

- **Iatrogenic iron overload** is a consequence of such frequent transfusions, and needs to be anticipated and prevented with chelation.
- **Chelation therapy** should commence when the ferritin has reached > 1000 mg/l, usually around the 10th–12th transfusion. **Desferrioxamine** is the most commonly used agent. It is given as a subcutaneous infusion over an 8- to 12-h period 5–6 nights per week. The injection sites give rise to the depigmented patches on dark skin that are seen in most patients. Ask about compliance in adolescents. Side-effects include visual problems and sensorineural deafness, so vision and hearing should be tested before therapy and regularly thereafter.
- **Folic acid supplementation** often required, as increased erythropoiesis uses up folic acid.
- **Splenectomy** may be necessary in some patients with severe symptoms from hypersplenism. This will often reduce the transfusion requirement, but there is a high risk of bacterial sepsis.
- **Allogenic bone-marrow transplantation** is considered to be curative, and is being used with increasing success. However, it carries a significant morbidity and risk of mortality, so patients need to be carefully selected.
- Remember the importance of a multidisciplinary approach, including specialist nurse, haematologist, cardiologist, psychologist, genetics team with genetic counselling for the family.

Sickle cell disease

Like thalassaemia, sickle cell disease is commonly seen in the exam. Due to the multiple ways in which the condition can present (including as a 'paediatric emergency'), children and families are also asked to attend the history taking/management planning station. If you work in an area where sickle cell disease is rarely seen, then you need to read about and be very familiar with the condition.

Tip +

Afro-Caribbean child with pallor, jaundice and no palpable spleen = sickle cell disease until proven otherwise

Instructions to candidate

Examine this 11-year-old child; what do you find?

Case

Jimmy is an Afro-Caribbean child who looks well. He does look short for his age and I would like to plot him on a centile chart. On close inspection of his sclerae he looks jaundiced. His conjunctivae look pale. He has what I think is a cholecystectomy scar, but the rest of his abdominal examination is normal. In particular, I cannot feel his spleen ...

Why can you not feel his spleen?

This child presents acutely unwell with a pyrexia, how would you manage him?

This child presents in acute respiratory distress, what are the possibilities and how would you manage him?

Clinical findings

- ❍ Typically Afro-Caribbean child, usually appears well. May have short stature
- ❍ Jaundiced (mild), pallor (seen as pale conjunctivae)
- ❍ May be cholecystectomy scar (increasingly laparoscopic)
- ❍ Splenomegaly in an infant, spleen not usually palpable in an older child
- ❍ Otherwise examination unremarkable

If acutely unwell look for

- Signs of infection
- Signs of dehydration
- Chest – sequestration
- Abdominal – acute splenic enlargement
- Joints – dactylitis

Notes on sickle cell disease

Sickle cell disease is caused by a point mutation in the beta-globin gene causing deoxygenated HbS to polymerise and distort the shape of the red blood cell, decreasing red cell lifespan. The red cell membrane damage that occurs leads to a haemolytic tendency. Sickled cells tend to sludge in the microcirculation, leading to vaso-occlusive syndromes. There are many genotypes, the most severe being HbSS.

It is most common in Africa, the Mediterranean, the Middle East and India.

In the UK, the incidence is around 1 in 2000 live births. Most new cases in the UK are now diagnosed on newborn blood spot screening.

If a neonate has HbSS, they will have 10% HbS, the rest being HbF. There are two different tests used for Hb analysis; if the first is positive then the alternative test is used to confirm. As in all good screening programmes, a positive result triggers a pathway of confirmatory testing and referral for follow-up.

The diagnosis in older children (eg those born in countries without a screening programme who have immigrated) is confirmed by haemoglobin estimation, blood film (contains target cells, poikilocytes and irreversibly sickled cells) and haemoglobin electrophoresis, which shows > 80% HbS.

Main clinical problems and their management

Usually asymptomatic until 5–6 months, when production of haemoglobin starts to switch from HbF to HbA. The problems that follow are due either to haemolysis or more frequently, vaso-occlusion – this inevitably involves the spleen which renders the child functionally asplenic:

- **Chronic anaemia** – with typical symptoms of lethargy, reduced exercise tolerance, headaches. Transfusions are limited to those patients with disabling chronic pain.
- **Jaundice** – raised unconjugated bilirubin due to increased red cell breakdown.

- **Aplastic crisis.** In common with other patients with chronic haemolysis, patients with sickle cell disease are at risk of developing an aplastic crisis secondary to parvovirus infection. Transfusion is warranted in some instances.
- **Recurrent acute painful crisis:** this is the most common symptom of sickle cell disease. Can be severe, sometimes requiring hospitalisation. The quoted mean incidence is one episode per patient-year. This obviously varies, with some patients having more episodes and some never experiencing pain. Dactylitis (hand/foot syndrome) is usually the first presentation in young children and can usually be managed in the community with fluids and analgesia. Abdominal pain is common, as is bone pain. Treatment: warmth, hydration, oxygen, powerful analgesia given promptly (eg intranasal diamorphine is easy to administer and quick-acting).
- **Splenic sequestration:** young children (particularly < 5 years) may have splenic enlargement associated with haemolysis and progress to hypersplenism. Blood pools in the spleen and this causes circulatory collapse, which may require life-saving blood transfusions. Parents should be taught to palpate their child's abdomen for any obvious splenic enlargement.
- Whether patients have splenic enlargement or autosplenectomy (from multiple infarcts), they all have **functional asplenia** by around 1 year of age and are therefore at risk of overwhelming infections from encapsulated organisms such as *Pneumococcus*. Patients must maintain full immunisation status and be on prophylactic penicillin from at least 3 months. They need prompt help if they become febrile with parenteral antibiotics.
- **Acute chest syndrome** describes a child with sickle cell disease who presents with an acute respiratory illness with new chest X-ray findings. Lung infarction/infection, or both, leads to symptoms not dissimilar to those of a severe pneumonia such as fever, cough, dyspnoea. All children will have an acute fall in Hb. Some children will have circulatory compromise and chest pain, with hypoxia exacerbating the problem.
- **Stroke:** occurs in 5%–10%. Can be present with hemiplegia and cranial nerve involvement. An urgent exchange transfusion is needed along with other management strategies for vaso-occlusive crises. Annual screening of Transcranial Dopplers can highlight those children most at risk, with this group needing more frequent transfusions to keep their HbS below 30% and decrease their risk of subsequent stroke.
- **Other problems:** Cholelithiasis (cholecystectomy may be required), renal papillary necrosis, avascular necrosis of femoral head, proliferative retinopathy, leg ulcers, priapism.

General management points

- Open access set up for patients along with a protocol for accident and emergency staff concerning the risks of sepsis/sickle crisis for any given sufferer of the disease. Particularly relevant in regions where the condition is rarely seen
- Manage acute vaso-occlusive crises: admit, warmth, oxygen, hydration, iv analgesia, iv antibiotics if indicated
- Use hydroxyurea in children who have reduced quality of life due to painful vaso-occlusive crises. The HbF fraction is increased on this treatment, leading in many to reduction of hospital admissions, as complications are rare if HbS is less than 30%
- Recognise and manage functional asplenia: immunise, iv antibiotics if evidence of infection and unwell
- Top-up/exchange transfusions mainly reserved for emergencies such as acute chest syndrome or stroke, or if quality of life becoming severely impaired due to recurrent acute crises
- Genetic counselling
- Multidisciplinary approach in community (including liaison with school) and in hospital
- Sibling allogenic bone-marrow transplantation is considered in selected severe cases where donor available

Variants

HbSC disease is a milder variant with moderately severe anaemia and splenomegaly. Vaso-occlusive episodes are less frequent and usually milder. Aseptic necrosis of the femoral head is common.

12.2.5 Disorders of haemostasis – haemophilia and von Willebrand's disease

Tip +

In a child with bleeding gums from toothbrushing, or dental/surgical bleeding, think about platelet deficiency or von Willebrand's factor deficiency

If there is prolonged bleeding or spontaneous bleeding, think about haemophilia

Haemophilia

The haemophilias are the commonest and the most serious of the congenital coagulation disorders. About 80% of cases are **X-linked haemophilia A**, causing a majority to have reduced factor VIII activity and some patients to have factor VIII antigen. It is recommended that you read a recent review article on the subject, as there are many aspects that could be discussed.

Notes on haemophilia A

Incidence is around 1 in 12 000 boys.

Numerous mutations in gene structure have been described. In 2/3 of cases the family history is positive. The degree of severity is similar among family members. If there is a known family history and a male baby is born, then the factor VIII level can be tested on cord blood. The severity of disease depends on the level of factor VIII activity in the plasma:

- Severe cases have < 1% of normal activity
- Moderate cases have 1%–5%
- Mild cases have 6%–30%

NB Carrier status can be tested and DNA markers identified, but only in specific situations and after referral for genetic counselling.

You may be asked about the typical results from a coagulation screen; the factor VIII deficiency causes a greatly prolonged activated partial thromboplastin time (APTT), with a normal platelet count, bleeding time and prothrombin time.

Clinical presentation

If the disease is not suspected on basis of family history, infants may present having developed severe bruising at birth or after immunisations. The commonest time of presentation otherwise is when crawling begins.

The hallmark is haemarthroses; knees, ankles and elbows are the most commonly affected joints. The bleeding itself occurs from the vascular synovium. The joint feels warm and will be swollen. Repeated haemorrhages may cause degenerative changes with osteoporosis, muscle atrophy and eventually a fixed immobile joint.

Bleeds into the GI tract, genitourinary tract, tongue and particularly brain can cause significant problems.

Management

Management of haemophilia and other bleeding disorders requires multidisciplinary expertise, co-ordinated from designated haemophilia centres. Children with mild to moderate haemophilia, may only require the administration of vasopressin (DDAVP), which releases endogenous factor VIII from its stores. Tranexamic acid tablets can be used for minor mouth bleeds. NSAIDS need to be avoided but paracetomol is safe.

The mainstay of medical treatment for those with severe haemophilia A, is the administration of recombinant factor VIII concentrate. This is given as regular prophylaxis against haemorrhage (aiming to get over 2% activity) after the first joint bleeds in toddlers, or as required for acute bleeding; up to 100% activity is needed when treating an intracranial haemorrhage.

Blood-borne diseases, particularly HIV, hepatitis D and hepatitis C, used to be a risk in the past before availability of recombinant factor VIII. One risk that remains is the development of factor VIII inhibitors. These render factor VIII replacement therapy less effective through antibody neutralisation and require progressively higher doses to be given; other agents such as recombinant factor VIIa often need to be given instead. This affects around 15% of patients with haemophilia A requiring factor VII concentrate. Children are screened for inhibitors on a regular basis.

Physiotherapy is important in patients who are recovering from joint bleeds as muscles will otherwise atrophy through lack of use.

Haemophilia B exhibits varied genetics and is associated with low factor IX levels. Because factor IX levels are normally low in neonates, it can be difficult to diagnose in patients under 6 months. Treatment is with recombinant factor. There is a 3% chance of improvement at puberty.

Notes on von Willebrand's disease

Von Willebrand's disease is the commonest inheritable bleeding disorder, and occurs when vWF synthesis is reduced or when abnormal vWF is produced.

Von Willebrand factor (vWF) acts as a carrier protein to protect circulating factor VIII from enzymes. It also mediates platelet adhesion.

Platelet *function* is therefore impaired and a child with this condition has an increased disposition to bleeding. Estimated prevalence is in the region of 1%. The gene for vWF is on chromosome 12p. If suspected, specific tests are needed to elicit quantity and function of vWF and factor VIIIc.

Unless severe, the majority of patients are asymptomatic, unless they are having surgery. Prolonged menorrhagia in older girls should also prompt suspicion. There are three types:

- Type 1: mild decrease in vWF levels and activity. Nosebleeds and easy bruising commonest presentations if symptomatic. Treat bleeding with DDAVP (watch sodium and fluid balance). Tranexamic acid can help symptomatically.
- Type 2: normal or slightly low amounts of vWF but abnormal vWF function. DDAVP (usually with tranexamic acid) can be effective for troublesome mucosal bleeds.
- Type 3: severe disease, complete absence of vWF. Often presents in neonatal period, eg large cephalohaematoma. Later on, haemarthrosis and muscle bleeds can occur. DDAVP ineffective.

For significant acute bleeds or anticipated surgical bleeds (particularly in types 2 or type 3, but can occur in type 1 too), replacement using vWF concentrate is required.

Management points to consider for patients with a severe bleeding disorder

- If antenatal diagnosis: plan delivery (no forceps/ventouse)
- Avoid intramuscular injections, aspirin, NSAIDs
- Subcutaneous hepatitis A and B vaccines (because of risks associated with frequent blood products)
- Inform GP and dentist
- Multidisciplinary team management
- Educate parents
- Think about preparing action plan for management of complications (who to ring, what blood products to give)
- Physiotherapy, eg for joint bleeds
- Genetic counselling
- Involve haematologist for treatment of acute events

12.3 Dermatology

12.3.1 Introduction

Skin problems may be seen as an 'other' case, often as part of a systemic condition. Café-au-lait spots in a child with neurofibromatosis would be one such example. Remember that the stations are 9 minutes long, so a 'spot diagnosis' of a lesion in a child with no other signs is going to be unlikely. Therefore it is worth practising a thorough examination of the skin. It is well worth attending a paediatric dermatology clinic, which will enable you to gain experience not only in recognising and describing common lesions but also in discussing their management.

The candidate should also review the appearance of common skin lesions in a dermatology atlas. Specific skin conditions are discussed in this chapter in note form to complement this revision.

12.3.2 Clinical examination

Brief scheme for examination of the skin

Remember to include a comment about the general appearance of the child (including their nutritional status, and the need to plot their height and weight on a centile chart) before you go on to assess the skin, hair, nails, teeth and mucous membranes:

- Aim for good exposure
- Look at the hands, nails, hair, teeth and mucous membranes
- Expose other areas and don't forget axillae and soles of feet
- If desperate, ask whether there is a rash and where it is
- Inspect, palpate and be prepared to describe the rash as you go along
- If you think a lesion is part of a systemic condition, ask to examine the relevant systems. Look for lymphadenopathy, hepatosplenomegaly and at the joints if appropriate
- Mention Wood's lamp examination if applicable; this will highlight areas of altered pigmentation such as vitiligo and certain types of fungal infection. It is also used to look for the depigmented patches associated with tuberous sclerosis

Notes on skin examination

When you have noticed a lesion and/or a rash, you will need to describe the appearance of the skin using the appropriate language.

Rash
- Distribution: remember to examine the palms, soles, mouth and scalp
- Pattern: symmetry, sun-exposed area, sites of pressure, etc
- Characteristic location: flexural, extensor surface, palms and soles, etc
- Extent of involvement: circumscribed, regional or generalised
- Configuration: dermatomal, linear, annular or grouped

Lesion (whether solitary or within rash)
- Type of lesion: macule, papule, nodule, vesicle, etc
- Shape of individual lesions: round, target, oval, linear, annular, etc
- Colour: red, brown, white, etc
- Blanching on pressure: present or absent
- Consistency of lesion on palpation: infiltrated, firm, soft, doughy, etc
- Tenderness

Glossary of lesions

Macule: flat lesion that differs from surrounding because of its colour; less than 5 mm in size, may have any shape

Patch: macule > 5 mm

Papule: small, solid, elevated (palpable), < 5 mm in diameter

Nodule: palpable, solid, round or ellipsoid lesion larger than papule (> 5 mm in diameter)

Plaque: raised, palpable lesion caused by thickening of the skin > 10 mm.

Petechiae: pinpoint (< 2 mm in diameter) haemorrhage in the skin, does not blanch with pressure

Purpura: larger than petechiae, does not blanch with pressure

Vesicle: circumscribed, elevated, blistering lesion that is < 5 mm in diameter, containing clear fluid

Bullae: vesicles > 5 mm in diameter

Pustule: circumscribed, raised lesion containing pus

Cyst: sac containing liquid, semi-solid material

Wheal: rounded, flat-topped papule or plaque caused by dermal oedema. Often has surrounding erythema or 'flare'

Scale: abnormal shedding or accumulation of stratum corneum in flakes, eg psoriasis

Crust: hardened deposits resulting from drying of exudates

Ulcer: defect in skin due to destruction of epidermis and part of dermis

Erosion: moist, circumscribed, depressed lesion due to loss of all or portion of viable epidermis

Scar: ulcer heals with a scar, as do chickenpox lesions in some children

Atrophy: a decrease in the size of a cell, tissue, organ or part of the body. Atrophic skin is thin and easily traumatised

Table 12.1: Useful definitions

12.3.3 Skin conditions commonly seen in the exam

- ❍ Eczema
- ❍ Psoriasis
- ❍ Mongolian blue spot
- ❍ Strawberry naevus
- ❍ Sebaceous naevus
- ❍ Lipodystrophies
- ❍ Vitiligo
- ❍ Molluscum contagiosum
- ❍ Ichthyosis
- ❍ Ectodermal dysplasia
- ❍ Epidermolysis bullosa
- ❍ Erythema multiforme

Discussed in Chapter 11:

- ❍ Capillary haemangioma (Sturge Weber) – Section 11.5
- ❍ Café-au-lait spots (neurofibromatosis) – Section 11.5

Eczema

Eczema is a chronic, recurrent, genetically influenced skin disorder of early infancy, childhood and adult life. Cases are common and there is much to talk about; children are used for both the clinical station and the history taking/management planning station. The distribution and morphology of lesions are diagnostic; there is no definitive test:

- ❍ Common problem – prevalence around 15% between birth and 12 years
- ❍ Multifactorial and associated with asthma/rhinitis. Family history of atopy in 70%
- ❍ Slight female preponderance
- ❍ Begins < 2 years in 70% of cases, and around 2/3 have long-term remission by 16 years
- ❍ Early-onset, severe disease carries a poorer prognosis.

There are characteristic patterns of evolution:

- ❍ **Infantile phase** begins at between 1 and 6 months and lasts for 2–3 years. Rash composed of red, itchy papules and plaques, many of which ooze and crust. Symmetrically distributed over the cheeks, forehead, scalp, trunk and extensor surfaces. Nappy area usually spared
- ❍ **Childhood phase** between 4 and 10 years. Symmetrical distribution over wrists, ankles, flexural surfaces of arms, legs. Circumscribed, erythematous, scaly, lichenified plaques. May get secondarily infected. Another feature in this age group is cheilitis; children can delevop very sore lips and perioral area. Some children develop additional folds of skin below the eyes called Dennie–Morgan lines, a sign of ongoing inflammation and oedema

- **Adult phase** mainly involves flexural creases of arms, legs, neck and hands/feet

In nearly all cases the skin is dry, even when it is not inflamed. Eczema can have a significant impact on the quality of life for the child and carers. In many cases chronic pruritus leads to sleep loss and emotional upset.

Treatment regimens can be time consuming and poorly tolerated by young children.

Eczema can also be secondarily infected with staphylococci, streptococci and Herpes simplex (eczema herpeticum). Children can become systemically unwell and require urgent treatment, especially if they have eczema herpeticum, which can be life-threatening.

Treatment

NICE has guidelines on the management of eczema in children under 12 years of age (www.nice.org.uk/cg57):

- Education of parents and patients, especially in the recognition of bacterially infected eczema and eczema herpeticum
- Avoidance of exacerbating factors and allergens
- Nail care – keep nails short to lessen trauma from scratching
- Emollients – emulsifying ointment, creams, bath oil
- Topical steroids, (escalating potency as required) and in some cases calcineurin inhibitors (eg tacrolimus)
- Occlusive bandaging
- Phototherapy
- Antibiotics or antiviral drugs for secondary infection

You may be asked to give a differential diagnosis.

Differential diagnosis of eczema

- **Contact dermatitis** – often clearly demarcated and linked to a known irritant
- **Hyper IgE (Job) syndrome** – levels of IgE extremely high: over 2000 IU/ml, and up to 50 000 IU/ml in some patients. Patients have recurrent boils, chest infections and sinusitis. Some have characteristically coarse facies with a prominent supraorbital ridge
- **Wiskott–Aldrich syndrome** – X-linked-recessive condition with thrombocytopenia and recurrent infections
- **Seborrheic dermatitis** – infant usually < 3 months with yellow scales on scalp, forehead, nappy area. Caused by a yeast called Malassezia

Psoriasis

- Common disorder, prevalence 1%–3%. Multifactorial – both hereditary and environmental factors involved.
- Around a third of patients develop the rash before 20 years of age.
- Characterised by red, well-demarcated plaques with a dry, thick, silvery scale. Distribution is over scalp, sacrum and extensor surfaces of extremities.
- In children, 50% have large plaques over knees, elbows.
- Can present as a persistent nappy-area dermatitis in infancy.
- Plaque psoriasis is the commonest subtype. There are other forms. Guttate psoriasis features drop-like lesions scattered all over the body, including the face, trunk, and extremities in one-third of cases. Often precipitated by streptococcal or viral infections. Erythrodermic and pustular psoriasis are rarer, the pustular form being a dematological emergency.
- Nail involvement in some, look for pitting and onycholysis (detachment of nail from its bed).
- Arthropathy is rare in children but should be looked for.
- Seen more frequently in patients with inflammatory bowel disease.

The diagnosis is clinical but can be confirmed on skin biopsy.

Particular signs that may be seen are:

- Koebner phenomenon – lesions induced in areas of local injury
- Auspitz sign – removal of scale causes multiple, small bleeding points

Treatment: this can involve emollients, topical tar, keratolytics and corticosteroids. Chronic lesions may require light therapy with UVB.

Mongolian blue spot

You may be asked to comment on purplish pigmentation over the lower back of a baby either after a full skin examination or as part of another examination. The differential is bruising and the possibility of non-accidental injury may have been raised:

- Bluish-black macules over lumbosacral area and buttocks of many Afro-Caribbean and Asian babies
- Due to concentrated collection of melanocytes deep in the dermis; fade away gradually and are rarely seen in children over 4 years of age
- Mistaken for bruising/non-accidental injury – so it is important to document them on baby/check notes if a child has been assessed in hospital for any reason. Otherwise they are of no clinical significance

Strawberry naevus (infantile haemangioma)

Instructions to candidate

Examine this girl's skin and tell me what you find.

Case

Leanne is a 3-month-old who looks well and is comfortable in her mother's arms …

You systematically examine skin as previously described.

… on full examination of her skin she has a prominent, bright red, protuberant lesion on the upper abdomen. It is well demarcated; I would like to measure it.

*I think this is an **infantile haemangioma,** also known as a strawberry birthmark.*

Questions

○ Will it get bigger? Will it disappear?
○ What are the potential complications?

Notes on infantile haemangioma

○ Common abnormality that can cause much anxiety
○ It is a benign vascular tumour and is normally bright red, protuberant, compressible and sharply demarcated
○ Usually not present at birth but usually appears within 1–2 months
○ More common in girls than boys, and in ex-premature than in term babies
○ Common sites – face, scalp, back, anterior chest – may be multiple
○ Develops in phases – rapid expansion (during first year), stationary (6–12 months) and spontaneous involution (develops blanched, pale, grey areas)
○ By 5 years, 50% involute; 90% by 9 years
○ In general no treatment is required and the parents are reassured in view of good outcome
○ Complications – ulceration, infection, haemorrhage, platelet consumption. May interfere with vision (interfering with the development of binocular vision), urination (urethra), airway (tracheo-bronchial). Visceral involvement (especially of the liver) can be fatal if untreated
○ Treatment is sometimes needed for very large lesions, or for lesions in which complications (especially recurrent bleeding and infections) have occurred. The options can include β-blockers, eg propranolol can be used to good effect as part of a management protocol, as their use needs to be closely monitored. Bronchospasm, diarrhoea and hypoglycaemia can occur,

especially in younger infants. Excision laser therapy rarely used apart from significantly ulcerated lesions or significant telangectasia. Corticosteroids if contraindication to β-blockers, or for life-threatening lesions. Interferon alpha for life-threatening lesions

You may be asked about the **Kasabach–Merritt syndrome**, which describes rapidly enlarging haemangiomas and leads to consumption of platelets (thrombocytopenia), clotting factors (coagulopathy) and red blood cells (microangiopathic haemolytic anaemia).

Sebaceous naevus

Again you may be asked to comment on this at the end of another case:

- Solitary, oval or linear, yellow-orange, warty lesion on the scalp
- Circumscribed area of hair loss
- Treatment – surgical excision
- Risk of neoplastic change (basal cell carcinoma) in 10%–15% cases after puberty

Lipodystrophy

Lipodystrophy is a loss of subcutaneous fat. You may see a child with either generalised or partial lipodystrophy. The clue is the prominent muscles with reduced subcutaneous fat, particularly over the face. It is important to have some knowledge of the causes and types of lipodystrophy.

Generalised lipodystrophy

- Congenital (autosomal recessive) or acquired (sporadic)
- Tall children, advanced skeletal maturation, prominent muscles, enlargement of penis/clitoris, abundant curly scalp hair
- Acanthosis nigricans, hirsutism, hepatomegaly, insulin-resistant diabetes
- May have renal, cardiac, neurological abnormality, seizures, developmental delay, carbohydrate intolerance, hyperinsulinaemia, hypertriglyceridaemia
- Growth hormone low despite tall stature

Partial lipodystrophy

- Girls > boys (4:1)
- Develops over a few months to several years
- Symmetrical subcutaneous fat loss initially over the face, upper trunks and arms – leads to disproportion between the upper and lower limbs
- Chronic glomerulonephritis associated in 25% of cases with low C3 complement due to activation of alternative pathway
- No treatment, prognosis is guarded

Tip +

Remember that in diabetes lipoatrophy and lipohypertrophy can occur at injection sites

Vitiligo

- Symmetrical, ivory-white depigmented patches
- Common problem; higher incidence in pigmented population. Half present under 20 years of age
- Associated with autoimmune diseases (thyroid, diabetes, pernicious anaemia, Addison's disease)
- Slow progression, static after variable period of time
- Spontaneous re-pigmentation in 10%–20% of cases
- Camouflage make-up can be used

Molluscum contagiosum

- Smooth, pearly papules with characteristic central punctum
- Caused by pox virus
- Occurs in crops, spreads by auto-inoculation
- Mostly no treatment is offered and the condition improves over 12–18 months
- Cryotherapy with liquid nitrogen and curettage have been used

Ichthyosis

Ichthyosis is characterised by dry, thickened skin, and the condition varies in severity. It must be distinguished from eczema, in which inflammatory changes play a key role. There are various specific syndromes, all of which are inherited disorders of keratinisation.

Types

- Ichthyosis vulgaris – autosomal dominant (most common, 1 in 250 and least severe)
- X-linked-recessive ichthyosis
- Lameller ichthyosis – autosomal dominant or recessive
- Bullous ichthyosiform erythroderma – autosomal dominant
- Non-bullous ichthyosiform erythroderma – autosomal recessive

These are all lifelong disorders with little spontaneous remission. Lamellar and ichthyosiform erythroderma both present at birth as a 'collodion baby' (encased at birth in shiny, tight membrane); other types present within 3 months of birth.

Severe problems from the outset: thick scales, corneal dystrophy, ectropion, fever and faltering growth. Treatment is with emollients, keratolytics, oral retinoids (ichthyosiform erythrodermas), antibiotics for secondary infection.

Multiple associated syndromes; for example, Netherton syndrome (autosomal recessive).

Ectodermal dysplasia

Instructions to candidate

Comment on this child's general appearance.

Case

> *Simon is a well-looking child with coarse wispy hair and sparse eyebrows and eyelashes. His skin appears to be dry. He has periorbital hyperpigmentation. He has pegged teeth (or false teeth).*
>
> *This would fit in with ectodermal dysplasia.*

Notes on ectodermal dysplasia

- The ectodermal dysplasias are a heterogeneous group of inherited conditions characterised by a primary defect in one or more of the following: teeth, nails, hair, other ectodermal tissue (eg eyes, ears, oral/nasal mucosa) and sweat gland function. Melanocytes and the central nervous system can also be affected.
- There are multiple gene defects which all lead to the abnormal development of fetal ectodermal structures. The most common form is X-linked recessive.

On examination (diverse features):

- Nails – thick, dystrophic and fragile
- Skin – atrophic, dry, wrinkled; requires emollients
- Scalp hair very sparse as there is a reduction in the number of hair follicles; wigs are often worn
- Facies – thick lips, saddle nose, frontal bossing, maxillary hypoplasia, cleft lip, cleft palate, abnormal ears
- Eye – dry eyes, hypoplasia of nasolacrimal duct may lead to corneal ulcers
- Teeth – reduced number of conical teeth, with the need for early use of prostheses
- Mucosa – dry mouth, thick nasal secretions, atrophic rhinitis

Clinically, mucosal involvement also leads to recurrent chest infections, otitis media and dysphagia. Asthma and rhinitis can be associated with this condition.

Faltering growth is common. Always offer to plot the height and weight if such a patient presents in the exam.

One life-threatening complication is hypohidrosis leading to hyperthermia; advice on cooling, activity, clothing, even relocation to cooler areas is important.

Management

- Precautions/advice re: potential for hyperthermia
- Early dental treatment
- Ophthalmological review: artificial tears are often needed
- Audiology review: hearing impairment is common
- Wigs
- Psychological input may be needed

Epidermolysis bullosa

- Mixed group of hereditary, bullous diseases characterised by the development (in response to trauma) of vesicles, bullae or erosions.
- Over 15 types based on inheritance, clinical features, electron microscopic identification of cleavage plane of blister:
    - Epidermolytic – within epidermis
    - Junctional – at the dermoepidermal junction in the lamina lucida
    - Dermatolytic – upper dermis

Best described as three major categories:

- Epidermolysis bullosa simplex (EBS) – intra-epidermal. There is a non-scarring (autosomal-dominant) form that presents at birth, and mucous membranes may be involved. Feet and hand lesions are common but may be generalised. Usually improves with time. There is also a localised type of EBS – also autosomal dominant and confined to hands/feet. Usually presents in the first 2 years of life. May be confused with friction blister
- Junctional epidermolysis bullosa (JEB) – autosomal recessive, presents at birth or shortly thereafter, often progressive. There are lethal and non-lethal types. Nails are lost, teeth are dysplastic. Large granulomatous ulcers are seen in the perioral area. Mucosal involvement may lead to oesophageal strictures. Anaemia and secondary infection are common complications
- Dystrophic epidermolysis bullosa (DEB) – autosomal dominant and autosomal recessive. Recessive form is the most severe, with devastating effects. Usually present at birth. Entire skin may be affected by minor trauma. Scarring, milia formation and nail loss occur. Oral involvement with

scarring of tongue and oesophageal stricture seen. Mitten-like hands/feet, contracture of large joints often result. Complications: infection, anaemia, growth retardation, amyloidosis, skin cancer (squamous-cell carcinoma), etc. Carries a poor prognosis

Management

- Supportive – nutrition, transfusion, gentle handling to minimise the likelihood of trauma, aseptic wound care and non-adherent dressing of lesions, antibiotics for infection, appropriate clothing and footwear
- Multidisciplinary involvement
- Genetic counselling

Erythema multiforme

This is an acute disorder, which starts off as red papules that may blister. They then form target lesions, so called because the appearance of a bruise in the centre gives them a target-like appearance.

If there is a patient with such a condition on the ward or who has been recently discharged, they may be on a shortlist of reserve patients to cover a potential shortfall in cases. This is a condition that is frequently seen in paediatric assessment/ambulatory units.

You will be asked about causes:

- Infections – Herpes simplex, *Mycoplasma*, Epstein–Barr virus, among others
- Drugs – penicillins
- Malignancy
- Systemic lupus erythematosus

Management

- Treat underlying cause
- Review if any signs of Stevens–Johnson syndrome, the severest end of the spectrum of this illness

Stevens–Johnson syndrome

There is often a prodromal illness. Patients develop severe erosions of at least two mucosal surfaces. Look for genital involvement, areas of skin necrosis (often on lips and mouth, leading to difficulties taking oral fluids), conjunctivitis. Patients can be very unwell and exudative fluid losses are high, much the same as in extensive burns. Such children need to be managed in a high dependency unit setting. Systemic antibiotics need to be given if secondary infection is suspected.

12.4 Endocrinology and growth

12.4.1 Introduction

An assessment or comment on growth is a part of almost all of the short cases in the exam. During the short cases, if you think a child is tall or short you should say so and say that you would plot them on a growth chart. Children with tall stature, short stature and conditions known to affect growth are frequently used as short cases (see Table 12.2). You will not be expected to know the details of the management of all of these conditions, however you will be expected to understand the principles on which this is based. You will need to have a structured approach to the assessment, investigation and management of the following conditions.

Familial short stature

Familial tall stature

Growth hormone deficiency

Turner syndrome

Noonan syndrome

Achondroplasia

Systemic disease affecting growth (eg cystic fibrosis, juvenile idiopathic arthritis, inflammatory bowel disease)

Storage disorders (eg Hurler syndrome)

Marfan syndrome

Klinefelter syndrome

Homocystinuria

Goitre/Hypo/Hyperthyroidism

Cushing syndrome

Table 12.2: Conditions that are seen commonly in the exam

12.4.2 Assessment of growth

Growth charts

World Health Organization (WHO) growth charts

In 2007 the growth charts used in UK were changed after a 15-year process involving data collection from birth to age 5 years in six different countries. The new WHO growth charts (for children from 0 to 4 years of age) were published in 2006 and adopted in the UK in 2007.

There is an excellent training package available on the website of the Royal College of Paediatrics and Child Health (RCPCH) (www.rcpch. ac.uk/Research/UK-WHO-Growth-Charts), which will provide you with a good knowledge of how to use these charts, as well as giving you PDFs of the different growth charts in use, including the ones used in the Personal Child Health Record. It is worth spending time familiarising yourself with these, as you may be asked to plot growth and you need to be able to do this quickly and confidently.

Reasons for the previous changes in the growth charts

- Healthy breastfed babies show similar growth patterns worldwide. Previous charts were based on a population of breast- and bottle-fed babies, and there are some differences in growth patterns between breast- and bottle-fed babies.
- Rather than describing average growth, it was felt that growth charts should reflect *optimal* growth. Therefore only data from healthy children were included. For all of these children the WHO feeding guidelines had been followed (ie they were breastfed exclusively until 4 months of age, with no solids before 6 months). All of the parents were non-smokers, and there were no economic barriers to providing normal nutrition.

Key changes seen in the WHO 2006 growth charts

- **A separate section for preterm births:** this is to be used for infants born between 32 and 37 weeks' gestation.
- **Average birth-weight centiles:** shown at 'age 0'–infants from 37 weeks' gestation onward are plotted at age 0 at birth (regardless of actual gestation).
- **No lines from birth until age 2 weeks:** this is to allow for variable amounts of weight gain and weight loss during this period. The presence of a gap emphasises the need to compare weights with birth weight, not centile lines, during this crucial period when feeding is becoming established.
- **De-emphasised 50th centile:** being above or below the middle line can be misinterpreted by parents, which is unhelpful. In the new charts the 50th centile line looks like all the others, which gives a clearer message about how growth charts are used.

- **Length/height discontinuity at age 2 years:** standard practice is to measure length, not height, until the age of 2 years. Length measurements tend to be slightly greater than height, which means that there is a small step down.

There is also guidance from the RCPCH on how to use their 2–18 years growth chart. This chart has a BMI plotting grid and a guide to pubertal assessment.

How to plot growth

The RCPCH guidelines suggest that you may be asked to plot growth during a short case. Be aware that in the clinical setting you/your consultants may have picked up time-saving bad habits when plotting a growth chart – in the exam it needs to be done by the book!

- Calculate age in weeks until 6 months, and thereafter calculate it in calendar months. To calculate the age in weeks you will need a date wheel or calendar. To calculate the age in months, use the birth date as a reference (eg if the child was born on 8 December then they will be 7 months old on 8 July of the following year).
- Make a dot (not a cross) in pencil (not in pen) to indicate the measurement. Do not join the dots.
- Preterm babies should be plotted on the preterm section of the 0–4 years growth chart if over 32 weeks' gestation, or on a separate preterm growth chart if born earlier than this. Once they reach a corrected gestational age of 42 weeks they are plotted on the normal growth chart (which starts at age 2 weeks). All babies born after 37 weeks' gestation should have their birth weight plotted at '0', and then start plotting again from age 2 weeks regardless of corrected gestational age.
- Once they are on the normal growth chart, plot their measurement by first putting a dot on their actual age, and then drawing a horizontal line with an arrow pointing to the spot which is their corrected age. This makes it clear to everyone who looks at the chart which measurement was the actual age and which was the corrected age.
- Continue plotting the corrected gestational age until age 1 year for preterm babies born at 32–36 weeks, and until age 2 years for babies born before 32 weeks.
- Children whose weight plots above the 99.6th centile should have their body mass index (BMI) calculated and plotted.

Suggested approach to interpreting growth charts

If you are shown a growth chart during the exam, you will be asked to provide an interpretation. By this the examiner is asking you to make observations about patterns of growth, and to relate them to a clinical context. There is a procedure for interpreting growth charts, and you need to practise this like any other clinical examination skill. A suggested approach is outlined below:

1. Check which chart has been used, and also that the correct gender chart has been used.

Growth charts available:

- Age-appropriate UK growth charts for boys and girls. There are '0–4' and '2–18' charts
- UK Growth Foundation charts (based on UK90 data) will still be in circulation for older children
- BMI chart
- Specialist growth charts for Down syndrome, Turner syndrome and achondroplasia
- Head circumference charts (from birth to 16 years)
- Growth velocity charts (which map centimetres of growth per year to age): peaks are seen in infancy and at puberty
- Childhood and Puberty Close Monitoring (CPCM) chart
- Preterm charts: these do not describe normal growth patterns of preterm babies, but are derived from birth-weight data at that gestation only. There is a UK WHO version of this available
- Sitting height charts and subischial leg length charts: these are based on Tanner–Whitehouse data

2. Comment on the current height and weight, mid-parental height and target centile range:

- Strictly speaking, if a measurement is within one-quarter of a centile on either side of the line, we say that it is 'on the centile'. For example, if a measurement is one-quarter either side of the 50th centile, the plot is 'on the 50th centile'. If the measurement is outside this, it is between centiles (eg between the 50th and 75th centile).
- This measurement should ideally be qualified by interpreting it within the context of expected growth. This is based on mid-parental height and target centile range. Traditionally these are marked in a small box on the right-hand side of the height chart.

It is worth familiarising yourself with mid-parental height calculator on the charts. Alternatively the mid-parental centile can be calculated by using the following formula:

Boy	*[(father's height in cm) + (mother's height in cm)]/2 + 7cm*
Girl	*[(father's height in cm) + (mother's height in cm)]/2 – 7cm*

- ○ Expected adult height centile usually falls within the target centile range which is 2 centiles above and 2 centiles below the mid-parental centile. However, this only applies if the parents have had a normal growth pattern themselves – beware in cases where there may be a familial abnormality of growth eg a skeletal dysplasia in one of the parents may 'normalise' abnomal growth in their offspring.
- ○ You may be expected to plot the parents' measurements. If these measurements are not available, you should mention that you need them to interpret the current height.
- ○ The target centile range is 8.5 cm on either side of the mid-parental height (this is approximately 2 centiles above and below the mid-parental height). If the child's measurement is outside this, refer to the child as 'short for family size' or 'tall for family size'.

3. Comment on the growth pattern:

- ○ It is essential to evaluate the weight in context with height, and if possible the trend in weight before commenting on linear growth. Look to see whether weight and height have always followed a particular centile. If not, at what age did the change occur? Is weight disproportionately affected compared with height, or vice versa?
- ○ Look for any sign of a pubertal growth spurt. If it occurs slightly earlier or slightly later than usual, it will look as if the height is crossing centiles upwards.

4. Comment on whether the growth is normal or abnormal.

You should always conclude your study of the growth chart by commenting whether it is a normal or abnormal growth pattern. When you are shown a chart in the short cases, the examiner will expect you not only to describe what you are seeing but also to interpret it in the light of what you already know about the patient. Normal patterns of growth are just as significant as abnormal growth patterns, because they may tell you that treatment is working or that certain diagnoses are unlikely.

Case – delayed puberty

I examined Katrina, who is a 16-year-old girl with some features that would be consistent with Turner syndrome. These were …

On review of her growth chart I see that she has always followed the 2nd centile for height and weight, and is short for family size based on her mid-parental height. I would like to take a full history and consider whether further investigations are merited (see 'Delayed puberty' in Section 12.4.9).

Case – Chronic disease

I examined Charlie, who is a 15 year old with some features that would be consistent with a diagnosis of cystic fibrosis. These were …

On review of his growth chart I see that he has an abnormal growth pattern, because he has always been short for family size, and his weight is frequently below the 0.4th centile. I would also like to plot his BMI on the appropriate chart. There is no evidence of a pubertal growth spurt on his growth chart, and he will need to have his puberty staged as part of his annual review. The most likely reason for this is that his underlying diagnosis of CF and poor nutrition is impairing his growth and delaying his puberty. I would also like to ask his parents at what age they went through puberty, to see if there is a family history of constitutional delay in growth and puberty. I would also like to dip his urine for glucose, to exclude cystic fibrosis-related diabetes mellitus.

Classic patterns of normal and abnormal growth

Normal growth in children can be divided into three phases. Though there are many factors which influence growth, each of these three phases have a different main influence:

- Infancy – nutrition
- Childhood – growth hormone and insulin-like growth factor (IGF-1)
- Puberty – sex steroids

Catch-up growth

This typically refers to infants who are born relatively undernourished (intrauterine growth retardation (IUGR)/small for gestational age), and who steadily increase through the centiles when given normal postnatal nutrition. Normally, if it is going to occur, catch-up growth takes place within the first few years of life. Be aware that catch-up growth in babies who were born small for gestational age may be a risk factor for cardiovascular disease later in life. Children who do not show catch-up growth by the age of 4 years (approximately 10%)

Catch-down growth

Some apparently healthy infants will cross down centiles without any obvious symptoms that are normally associated with faltering growth. These infants may be re-establishing themselves on to their optimum centile after a period of excessive weight gain in utero, such as in babies of diabetic mothers who are born macrosomic. On the growth chart you would expect to see a gradual slipping of the weight centile over the course of several weeks, finishing on a centile which is then followed normally. Before the expected centile is reached, this can be indistinguishable from faltering growth, and a careful history and clinical examination are important for avoiding undue concern and excessive investigation.

Faltering growth

This used to be known as 'failure to thrive'. This refers to weight crossing the centiles downwards in infancy. It also indicates the need to seek out an underlying cause, which could be related to nutrition, the environment, or an underlying disease or syndrome. Watch for faltering growth which occurs at weaning age or when formula is added – this could reflect compromised intake, food allergy, malabsorption or be an early marker of chronic disease/neurological problems.

Constitutional delay in growth and puberty

This is the commonest cause of delayed puberty. It presents to paediatricians most frequently in boys, but the incidence in boys and girls is probably similar. On the growth chart a slowed height velocity is seen just before puberty (this is normal), which leads to height crossing the centiles downwards. This becomes obvious at the age when most children are in their mid-pubertal growth spurt, which leads to an increasing gap in height between the patient and their classmates. It is commonly associated with a family history of pubertal delay (ask the mother at what age she started to menstruate, and ask the father whether he had his growth spurt around the same time as his peers). Investigations may show prepubertal (low) levels of luteinising hormone (LH) and follicle-stimulating hormone (FSH), and a delayed bone age. This normal growth pattern can be difficult to distinguish from pathological causes of poor growth. It is therefore important to make sure that there is not a chronic disease process, syndrome or endocrine cause (eg pituitary and thyroid disease, Cushing syndrome) contributing to persistently poor growth and failure to enter puberty. In girls, Turner syndrome needs to be specifically excluded by carrying out a karyotype.

Precocious puberty

Precocious puberty is defined in a girl as breast development before the age of 8 years, and in a boy as testicular volumes of 4 ml or more before the age of 9 years. On the growth chart this looks like a rapidly increasing height (crossing centiles upwards) at an age when the child should be prepubertal. These children may well be tall for family size.

Hypothyroidism

This typically causes gradual slowing of height velocity (height crossing down the centiles over a period of months to years) while the weight continues to follow or increase across centiles. This will result in an increased BMI and a relatively short and overweight child. After initiating treatment the height will increase rapidly, and BMI should fall.

Chronic disease

Many factors contribute to poor growth in children with chronic disease. Chronic disease is associated with cytokine release, which impairs normal bone growth mechanisms and responsiveness to endogenous GH and IGF-1. It can also result in relative undernutrition, particularly if requirements are high. Adequate nutrition is crucial for normal linear growth, from birth to final height. In patients who have chronic disease you would expect to see weight and height crossing the centiles downwards, with the child often being underweight for their height. Periods of disease exacerbation and/or poor compliance with medical treatment are often reflected in the growth chart, with poor growth during the period of active disease. In these patients the growth chart tells a story of disease severity and the impact of the disease, as well as indicating treatment success or failure. It is important to consider factors such as corticosteroid treatment (frequently used in children with chronic inflammatory conditions) which are likely to have an impact on growth.

Bone age

This involves a left wrist X-ray, which is interpreted based on which growth plates have fused. Interpretation of bone age is summarised in Table 12.3.

Delayed bone age	Advanced bone age
Increased cortisol	Increased androgens or oestrogens (precocious puberty)
Emotional deprivation	Hyperthyroidism
Decreased androgens or oestrogens (delayed puberty)	Cerebral gigantism (Sotos syndrome)
Poor nutrition	Weaver syndrome
Growth hormone deficiency	
Chronic disease	
Turner syndrome	
Constitutional delay of growth and puberty	

Table 12.3: Interpretation of bone age from left wrist X-ray

12.4.3 Tall stature

Suggested approach to tall stature examination

General approach

- Look at any growth charts in the room. Comment on the pattern of growth as well as the current centiles. Make a decision on whether the growth pattern appears 'within normal limits' or is 'not a normal growth pattern'. Remember that 1 in 250 normal children will fall outside the 0.4th and 99.6th centiles
- Enquire whether the parent's heights are available. If they are, calculate the mid-parental centile (MPC) and target centile range and make a decision about whether the child's growth is 'tall for family size'
- Look at the height velocity – as a general rule in endocrine causes of tall stature, the height velocity is excessive (as opposed to syndromic/obesity where height velocity is ≤75th centile)
- Look at the parents – do they look short/tall/dysmorphic? Comment on any obvious findings
- If a diagnosis becomes clear (for example, obvious thyroid eye disease), direct your examination towards confirming diagnosis and looking for complications

Instructions to candidate

This child has tall stature. What do you think is the cause of this? Look at this child and tell me what you observe?

Initial procedure

- Introduce yourself

General observations

- Well or unwell?
- Growth observations: height, weight, head circumference (offer to plot on growth chart)
- Obvious dysmorphic features, eg Marfanoid (direct further examination towards this)
- Any obvious thyroid signs? (direct examination towards this)

Arms

- Hands: hyperthyroid features (tremor/sweatiness), arachnodactyly, ligamentous laxity, ring and thumb sign (Marfan's, see below)
- Measure arm span and compare to height if possible
- Brachial pulses: compare the two sides. Assess character (aortic root dilatation), rate/rhythm (hyperthyroid)
- Blood pressure

Head and neck

- Macrocephaly (associated with Sotos syndrome)
- Mouth: high arched palate, dental crowding
- Thyroid eye signs
- Look for goitre or scar from surgery
- Listen for thyroid bruit
- Palpate neck for goitre

Chest

- Breast development in boys (obesity/Klinefelter)
- Signs of puberty (precocious puberty)
- Pectus excavatum, scars from surgery
- Café-au-lait spots (Marfan syndrome), any neurocutaneous markings (precocious puberty)
- Perform the cardiovascular exam now (if Marfan syndrome suspected)

Spine

- Examine for scoliosis (shoes off, pelvis straight, correct for postural elements, look for curvature standing and bending over) – a feature of Marfan syndrome but also seen in tall pubertal children
- Look from the side for kyphosis
- Look for ligamentous laxity, eg touch palms of hands to the floor while keeping legs straight

Also ...

- Always mention need to plot parental height/mid-parental centile
- Direct final examination towards most likely cause:
    - Mention cardiovascular exam/fundoscopy/arm span versus height if you suspect Marfan syndrome
    - Remember reflexes if thyroid signs are identified
    - Remember development/fundoscopy if homocystinuria is suspected
    - Mention pubertal staging if Kleinfelter syndrome is suspected

If asked for investigations you should mention:

- Karyotype
- Thyroid function
- Bone age
- Any others suggested by examination findings, such as IGF-1, sex steroids

Some causes of tall stature

- ○ Familial
- ○ Obesity
- ○ Syndromes:
  - Marfan
  - Homocystinuria
  - Sotos syndrome
  - Kleinfelter syndrome
  - Beckwith–Wiedemann (normal final height)
- ○ Endocrine
  - Hyperthyroidism eg Graves' disease
  - Precocious puberty:
    - a. central, eg brain tumour
    - b. peripheral, eg androgen mediated from congenital adrenal hyperplasia, adrenal tumour
  - Growth hormone excess – (extremely rare in paediatrics)

12.4.4 Conditions and cases associated with tall stature

Marfan syndrome

Marfan syndrome commonly comes up in the exam and you should be familiar with the clinical signs and management:

- ○ Prevalence of Marfan syndrome is 1:5000–10 000
- ○ Autosomal-dominant inheritance (20% de novo mutations)
- ○ Mutation in the fibrillin-1 gene on chromosome 15

Long-term complications are aortic dissection/regurgitation, kyphoscoliosis and lens dislocation. Children with Marfan syndrome will need yearly echocardiograms to compare their aortic root dimensions against centile charts, and regular ophthalmologic assessment. In the paediatric clinic it is important to monitor blood pressure regularly.

Examination findings

- ○ Tall for family size (but remember one of the parents may have Marfan syndrome, as it is autosomal dominant)
- ○ Long thin face, high arched palate, dental crowding
- ○ Arachnodactyly – long slender fingers (ask the patient to encircle opposite wrist with thumb and little finger)
- ○ Sternberg's sign – thumb adducts across palm

- ○ Ligamentous laxity – can place palms on the floor when the legs are straight, pes planus
- ○ Arm span greater than height
- ○ Pectus excavatum or carinatum
- ○ Scoliosis
- ○ Aortic root dilatation – examine brachial pulse character (compare both sides), praecordium for heave/thrill, apex beat character, presence of diastolic murmur
- ○ Mitral valve prolapse (late systolic murmur at apex)
- ○ Lens dislocation – up and in
- ○ Pes planus

Case – Marfan syndrome

I examined Jake, who is a 10-year-old boy who appears to be tall for his age. He is accompanied by his father who also appears tall. I would like to plot his height and weight on a centile chart. I would also like to calculate the mid-parental centile and target centile range.

He has features of Marfan syndrome. In particular he has evidence of ligamentous laxity, his arm span is greater than his height and he has a high arched palate with some dental crowding. There is no evidence of scoliosis, and his heart sounds and praecordial examination were normal. I would like to complete my examination by measuring his blood pressure in both arms, and looking at his eyes with an ophthalmoscope. I would also like to plot his mid-parental height on a centile chart.

Homocystinuria

- ○ Autosomal-recessive inheritance

Examination findings are similar to those in Marfan syndrome with some significant differences (see Table 12.4). Other important differences include psychiatric morbidity (50%), and increased risk of thromboembolic disease.

	Marfan	Homocystinuria
Joints	Hyperextensible	Contractures
Lens dislocation	Up and in	Down
Facies	Long	Ruddy complexion
Intelligence	Normal	Below average
Cardiac	Aortic root dilation	Normal
Bones	Normal	Osteoporosis
Urine	Normal	Excessive homocystine

Table 12.4: Differentiating between Marfan syndrome and homocystinuria

Klinefelter syndrome (47XXY)

- Incidence 1:1000–1500 males
- Individuals are tall with long limbs
- Usually presents as delayed puberty or gynaecomastia (present in 40%)
- Testicular volumes rarely exceed 6 ml
- Poor growth of pubic and facial hair
- Below average intelligence, psychosocial problems
- twentyfold increase in risk of breast cancer

XYY syndrome

Children present with above average height, and intelligence within normal limits, but an increased incidence of behavioural difficulties. They have normal genitalia and pubertal development.

Sotos syndrome (cerebral gigantism)

- Incidence 1:14 000
- Born large for gestational age (LGA) with macrocephaly
- Dysmorphic features include: hypertelorism, prominent forehead and chin, hands, feet, ears, nose and genitalia can be large
- Hypotonia +/– dyspraxia
- Bone age is usually advanced, so final adult height is usually normal

12.4.5 Short stature

Suggested approach to short stature examination

Instructions to candidate

Examine this child who is being investigated for short stature.
Look at this child. Tell me what you see.

Initial procedure

Initial general observations are vital to targeting your examination:

- Introduce yourself as previously described
- Look at any available growth charts
- Decide whether the child is short for family size or not
- Do they look well or unwell
- Any dysmorphic features?
- Underweight or overweight?
- Limb spine/disproportion?

The remainder of the examination can be targeted based on the observations above.

Short, underweight or ill child

Examination directed towards finding an underlying chronic disease process that may have impaired growth, for example:

- Cystic fibrosis/bronchiectasis
- Coeliac disease (wasted buttocks)
- Inflammatory bowel disease (mouth ulcers, nasogastric tube)
- Type 1 diabetes mellitus – poorly controlled diabetes will impair growth

Short, well and normal or overweight child

Look at growth pattern if growth chart available. Look for any evidence of an endocrine disorder:

- Cushingoid?
- Hypothyroidism? (examine for goitre)
- Hypopituitarism/growth hormone deficiency? (classically associated with central obesity and characteristic midfacial crowding – not always obvious)
- Remember the more significant the short stature (ie the further below the 2nd centile), the more likely it is that there will be underlying pathology. Overall though, the most common cause of short stature in a well child is familial short stature or constitutional delay in growth and puberty.

Summary

Finish examination by asking to plot parental heights and calculate mid-parental centile (MPC) and target centile range:

- Be prepared for the fact the child may be normal. Be prepared to discuss further investigations. These could be directed at looking for chronic disease (CRP/ESR, FBC, sweat test, coeliac screen, urine dip, etc) or endocrinopathy (thyroid function tests, insulin-like growth factor-1). Bone age is helpful in some circumstances.

Short, dysmorphic child

Examination should be directed to noticing patterns of dysmorphic features that might aid in diagnosing the reason for the short stature. If a 'spot diagnosis' is obvious or strongly suspected (eg achondroplasia, Turner syndrome), then direct examination towards identifying the associated physical characteristics and looking for complications. If there is no obvious diagnosis, then direct examination towards accurately describing what you see.

If something is obvious make sure you tell the examiner you have spotted it!

Growth

- Offer to measure height (including sitting height if you suspect a skeletal dysplasia), weight, head circumference and if appropriate, subscapular/triceps skinfold thickness and pubertal stage, and plot on appropriate centile charts. Use disease-specific growth charts where relevant
- You may be asked to plot height velocity on the relevant chart – practise this in clinic, and be quick, accurate and efficient in doing so
- If a short child is obviously post pubertal it is relevant, and should be stated

Is the child obviously dysmorphic?

- For example, Turner syndrome, Noonan syndrome, Russell–Silver syndrome, Prader–Willi syndrome, Cornelia de Lange syndrome, Rubenstein–Taybi, mucopolysaccharidoses. If so, then direct the examination towards that problem. Physical signs for the more commonly seen of these conditions can be found in the relevant 'Notes on short stature' section (page 408 onwards).

Limb/spine proportions

- Extend arms down lateral side: consider limb shortening if they do not reach the mid pelvis in infancy or the upper thigh after infancy. Consider spine shortening if arms extend well down upper thigh
- Examine arms for rhizomelic (proximal), mesomelic (distal) or symmetrical shortening. A useful way to do this is to fully flex the arm at the elbow and compare the upper arm with the lower arm
- Extend arm and examine carrying angle (cubitus valgus in Turner syndrome)
- Examine hands for hypoplastic, hyperconvex nails (Turner syndrome), short 4th metacarpal (Turner syndrome and pseudohypoparathyroidism type 1A)
- Look at spine for kyphosis/scoliosis. Take off their shoes and look from the side as well as from behind. Ask the child to bend over as part of the scoliosis examination. Look for any evidence of spinal surgery/back braces, etc
- Look at limbs for joint contractures/scars
- Measure and plot head circumference

Also I would like to …

- Look at parents' size (?dysmorphism/limb disproportion) and think about mid-parental centile/offering to plot parents' heights
- Where relevant don't forget development
- If dysmorphic features are found, remember that other systems, particularly cardiac, may be important
- Be prepared to discuss investigations. Genetic testing, white cell enzymes, bone age, skeletal survey are all useful for diagnosis depending on examination findings. Remember cardiac investigations, renal ultrasound, audiology, ophthalmology if looking for associated anomalies

Notes on short stature

Short stature is defined as a height less than 2 standard deviation score (SDS) below the mean (ie less than the 2nd centile). Short stature is a common reason for children to be referred to paediatricians. This is considered abnormal if a child is short for family size (ie outside the target centile range) or if there is an abnormal growth pattern such as crossing 2 centiles downwards. Normal growth requires adequate nutrition, wellbeing, normal endocrine system, the absence of chronic disease and normal bone metabolism.

Causes of short stature

- Familial short stature
- Constitutional delay of growth and puberty
- Short stature following being born IUGR:
  - Chronic disease process (short and thin)
- Endocrine (short and overweight):
  - growth hormone deficiency
  - hypothyroidism
- Cushing syndrome
- Syndromic: Turner syndrome, Noonan syndrome, Russell–Silver syndrome, Prader–Willi syndrome, Cornelia de Lange syndrome, (many others)
- Skeletal dysplasia: achondroplasia, hypochondroplasia (many others)
- Emotional deprivation/ neglect

Growth hormone deficiency

Affected individuals typically are short and have slowing of height velocity. They may have characteristic facial features (midfacial hypoplasia) and excess subcutaneous fat. IGF-1 can be a helpful screening test for growth hormone deficiency (ie if the IGF-1 is in the lower end of the reference range, growth hormone deficiency is more likely than if it is a robust level in the upper end of the reference range). Confirming the diagnosis requires a growth hormone stimulation test (eg glucagon, arginine, clonidine or insulin tolerance test). These should only be done in a specialist setting.

Proven growth hormone deficiency is an indication for recombinant growth hormone replacement. In the majority of cases this treatment will enable the child's final height to end up within the expected target centile range (depending on the age at which treatment is started).

Growth hormone replacement therapy

This involves a daily subcutaneous injection with recombinant growth hormone. The injection is given with a pen device similar to an insulin pen. There are six conditions for which there is a license to prescribe growth hormone and these indications are NICE approved (www.nice.org.uk/TA188) (see Box). With the exception of Prader–Willi syndrome, growth hormone treatment is used to increase a child's height and final height. The use of GH in Prader–Willi syndrome is to improve body composition and not to increase height (although this may occur during treatment). The dose of growth hormone varies depending on the indication for use.

Indications for growth hormone therapy, according to NICE guidelines

Growth hormone deficiency
Turner syndrome
SHOX deficiency
Prader–Willi syndrome
Chronic renal insufficiency
Born small for gestational age with no catch-up growth by age 4 yrs

The above indications may need to be discussed in more detail so are described below:

1. Growth hormone deficiency. Many children will stop taking growth hormone once final adult height has been attained.

There is some evidence to suggest that ongoing treatment improves the sense of wellbeing and quality of life of adults with growth hormone deficiency. It may also have a beneficial effect on body composition and cardiovascular risk factors.

2. Turner syndrome. Girls with Turner syndrome, although not growth hormone deficient, will reach a taller final adult height if given growth hormone. This needs to be started early for the best effect. Growth hormone will increase the final height by a mean of approximately 7 cm.
3. SHOX deficiency is an abbreviation for SHort stature hOmeoboX-containing gene. Approximately 15% of children presenting with short stature will have a heterozygous defect in this gene, which is present on both X and Y chromosomes. The clinical expression is very variable, but worth considering in a child with otherwise 'idiopathic short stature'.
4. Prader–Willi syndrome. Growth hormone therapy is given to improve body composition and increases muscle mass, strength and tone. Although height may improve, short stature is not a reason to initiate treatment. There have been concerns of worsening obstructive sleep apnoea while on growth hormone treatment, particularly in those who are obese, and most children with Prader–Willi syndrome should have a baseline sleep study before treatment, and a follow-up study while on treatment.
5. Chronic renal insufficiency. This is believed to cause a reduced sensitivity to growth hormone, so patients with this condition benefit from replacement therapy.
6. Small for gestational age. Approximately 10% of children fail to show any catch-up growth, and growth hormone is licensed for use if no catch-up growth is seen before the age of 4 years. Studies have shown a mean increase in final height of approximately 5–7 cm.

Several side-effects of growth hormone treatment have been reported, although these are rare. They include the following:

- Lipoatrophy or lipohypertrophy at the injection site
- Headache: this is normally transient
- Oedema in patients with Turner syndrome
- Hypothyroidism
- Scoliosis: this may occur in patients who were always going to have a degree of scoliosis, and may be precipitated earlier by the growth hormone treatment
- Arthralgia
- Slipped upper femoral epiphysis
- Insulin resistance: this can lead to worsening control in patients who already have diabetes. It is not thought to cause problems in non-diabetic patients on growth hormone treatment

- Theoretical risk of cancer: the reported occurrence is no higher than that in patients who are not on growth hormone treatment. This theoretical risk requires careful discussion with patients and parents.

12.4.6 Conditions and cases associated with short stature

Turner syndrome

- Occurs in 1:2500 female births
- 50% are due to 45XO, the remainder have other X chromosome abnormalities
- Variable phenotype (hypogonadism and short stature most consistent features)
- The diagnosis should be considered in all girls with growth or pubertal failure.

Features of Turner syndrome

General

- Short stature
- Webbed neck
- Wide-spaced nipples
- Cubitus valgus
- Chronic, severe otitis media

ardiac

- Bicuspid aortic valve
- Coarctation of the aorta
- Aortic dissection

Gonadal dysgenesis

- Streak ovaries
- Primary amenorrhoea
- Delayed puberty (15% will enter puberty spontaneously – but will not fully progress through puberty)

Renal and metabolic

- Increased risk of congenital renal abnormalities
- Hypertension
- Type 2 diabetes mellitus
- Hyperlipidaemia

Girls with Turner syndrome also have an increased incidence of learning difficulties and psychological/behavioural problems.

Management involves regular follow-up including:

1. Screening for congenital anomalies and associated complications
2. Growth hormone replacement therapy
3. Oestrogen/progesterone replacement in adolescence
4. Other important follow-up includes targeting educational needs (eg mathematical ability), screening for audiological complications (eg otitis media), hypertension, coeliac disease

Suggested examination structure for Turner syndrome

General

- ○ Plot growth (remember Turner syndrome growth chart)

Arms

- ○ Oedema of hands and feet in neonate
- ○ Hypoplastic and hyperconvex nails
- ○ Short 4th/5th metacarpals
- ○ Examine peripheral pulses
- ○ Widened carrying angle (cubitus valgus)

Head

- ○ Low posterior hairline
- ○ Prominent ears
- ○ Neck webbing
- ○ High arched palate
- ○ Look in ears (glue ear)

Chest

- ○ Shield chest
- ○ Widely spaced nipples
- ○ Scars from cardiac surgery
- ○ Multiple naevi
- ○ Breast development (think about pubertal stages and hormone supplements)

Also need to examine

- ○ Thyroid status (autoimmune hypothyroidism?)
- ○ Cardiovascular system (in particular four-limb blood pressure)
- ○ Pubertal staging

Case – Turner syndrome

> *Sarah is a 12-year-old girl who looks well. She appears short for her age and I would like to plot her height and weight on a centile chart. On general examination she has webbing of her neck and cubitus valgus. She has a short 4th metacarpal and dysplastic nails. The most likely diagnosis is Turner syndrome. I would like to examine her cardiovascular system and stage pubertal development.*

Noonan syndrome

- Autosomal-dominant inheritance
- Incidence 1:1000–2500 children
- Mild mental retardation in 30% (mean IQ – 85)
- Congenital heart disease (in 80%) – pulmonary stenosis, ASD, VSD, HOCM
- Coagulation defects in 30% (eg von Willebrand' disease, thrombocytopenia, clotting factor abnormalities, juvenile myleomonocytic leukaemia).

Examination findings

- Short stature
- Typical facies – hypertelorism, epicanthic folds, ptosis, micrognathia, low-set and posteriorly rotated ears, deep philtrum, small upturned nose
- Widely spaced nipples, cubitus valgus, short stature and pubertal delay
- Abnormalities on cardiac examination correlating to specific congenital heart disease
- Cryptorchidism, herniae

Case – Noonan syndrome

> *Jason is a 6-year-old boy who appears short for his age. I would like to plot his height and weight on the appropriate charts. He has hypertelorism, ptosis, micrognathia, epicanthic folds and low-set ears. Examination of his chest reveals widely spaced nipples and an ejection systolic murmur grade 3/6, loudest in the pulmonary area. It does not radiate to his carotid arteries. The most likely diagnosis is Noonan syndrome. His murmur is likely to be due to pulmonary stenosis. I would also like to examine his genitalia for cryptorchidism and stage his puberty.*

Russell–Silver syndrome

- Occurs in approximately 1:75 000 births
- Associated genetic abnormalities – maternal uniparental disomy of chromosome 7, and methylation defect in Beckwith–Wiedemann gene on chromosome 11

The syndrome is diagnosed clinically and should be considered if the following are present:

- Characteristic facial features (see below, 'Clinical features')
- Relative macrocephaly
- IUGR/low birth weight
- Short stature and poor postnatal growth

Clinical features

Head and neck

- Small, triangular face with a high forehead and micrognathic jaw
- Short stature and a normal head circumference (this gives the appearance of a large head, ie relative macrocephaly)
- Blue sclerae
- Prominent nasal bridge and downturned corners of mouth
- Late closure of the anterior fontanelle
- Sprengel's neck deformity (unilateral shortening and webbing to trunk)

Limbs

- Hemihypertrophy or limb asymmetry
- Clinodactyly
- Camptodactyly (fixed flexion of the fingers)
- Syndactyly of toes

Other

- The child may have hypospadias or undescended testes
- Feeding difficulties and hypoglycaemia (may require a gastrostomy)

Review of the child's growth chart is crucial for diagnosis

- Born small for gestational age
- Head circumference normal for age
- Little or no catch-up growth

The website of the Child Growth Foundation (www.childgrowthfoundation.org) is a good source of further information about Russell–Silver syndrome.

Mucopolysaccharidoses

Knowledge of the mucopolysaccharidoses and similar syndromes is important as these are common cases in the exam. Hurler and Hunter syndromes are the most frequently encountered.

Suggested examination structure for a child with mucopolysaccharidosis

General observations

- Comment on growth
- Describe coarse facial features
- Thickened lips
- Large head with frontal bossing
- Flat midface
- Prominent sutures
- Broad flat nose
- Comment on evidence of developmental problems (eg wheelchair)
- Look at dorsum of wrist for carpal tunnel surgery correction scar (common)

Head

- Examine corneas for clouding (appears after 1 year and not present in Hunter's)
- Examine fundi for cherry red spot (also present in GM1 gangliosidoses, which is a differential)
- Comment on nasal discharge (common and persistent)
- Listen for upper airway obstruction

Cardiovascular system

- Examine cardiovascular system for signs of congestive cardiac failure

Abdomen

- Note umbilical hernia if present
- Examine for hepatosplenomegaly

Finally

- Note joint stiffness and contractures
- Examine spine for exaggerated kyphosis (usually present), gibbus (the hump seen due to a deformed spine), scoliosis and cervical rod surgery (common in Maroteaux–Lamy and Morquio)
- Offer to perform developmental examination, and expect development to be delayed

Case – Mucopolysaccharidosis

> *I examined Jenny, who is a 5-year-old girl with some dysmorphic features. She has short stature, as evidenced by her growth chart. She also has macrocephaly and a short neck. There is some coarsening of her facial features; in particular she has thickened lips and some frontal bossing. She has very noisy breathing suggestive of upper airway obstruction. I suspect she has a mucopolysaccharidosis disorder. I would like to look in her eyes for corneal clouding and examine her abdomen for hernias and hepatosplenomegaly. I would also like to examine her cardiovascular system and her development, including hearing.*

Difference between **Hunter** and **Hurler syndromes**

	Hunter	Hurler
Inheritance	X-linked recessive	Autosomal recessive
Corneal clouding	No	Yes
Severity	Less	More
Nodules over scapulae	Yes	No

Table 12.5: Difference between Hunter and Hurler syndromes

Differential diagnosis

Other mucopolysaccharidoses:

- Morquio syndrome – very severe physical features identical to Hurler syndrome, but normal intelligence. Will have had cervical stabilisation surgery
- Maroteaux–Lamy syndrome – phenotypically similar to Morquio syndrome
- Sanfilippo syndrome – less severe physical features, but profound mental retardation

Skeletal dysplasia

- Around 200 disorders
- Include osteodysplasias and chondrodysplasias
- Chondrodysplasias are characterised by disproportionality of areas affected, eg mainly affecting limbs or mainly affecting trunk
- Diagnosis is based on skeletal survey and family history/genetics
- Some skeletal dysplasias have non-skeletal-associated anomalies (eg Ellis–van Creveld)

Case – skeletal dysplasia

> *Sarah is a well-looking girl who appears to be short for her age, as evidenced by her growth chart. She is post pubertal. There is no obvious limb disproportion but she has a marked kyphosis. I note that her mother has a similar body shape. I suspect she has a skeletal dysplasia and I would like to measure her sitting height.*

Achondroplasia

Incidence 1:20 000, autosomal dominant and is caused by mutations in the FGFR3 gene; more than 50% of cases are from sporadic mutations. Patients can expect to have a normal life-span but sudden death can occur due to cervical cord compression because of a small foramen magnum. Hydrocephalus may occur and may require shunting.

Height, weight, head circumference and development charts exist specifically for this condition. Spirometry should be performed as all patients have reduced lung capacity. Obstructive sleep apnoea may occur (consider organising a sleep study if symptomatic).

A key radiological finding is decreasing interpeduncular distances in lumbar spine (distinguishes this from hypochondroplasia).

Suggested examination structure for achondroplasia

If you are faced with a short child who obviously has achondroplasia, this is an approach to take in your examination.

General

- Growth
- Describe the facial features to the examiner: frontal bossing, flat nasal bridge, large head circumference, mandibular prognathism when older
- Look at parents and comment on whether they have achondroplasia

Limbs

- Hands are short and broad with a trident appearance
- Laxity of the ligaments
- Limbs show a rhizomelic (humerus/femur) shortening, progressive tibial bowing may require surgery

Spine

- Examine spine for gibbus in infancy, followed by lumbar lordosis and scoliosis in adolescence

Finally

- Examine the cardiovascular system for evidence of pulmonary hypertension, secondary to recurrent sleep apnoea
- Offer to examine the lower limb reflexes for evidence of spinal cord compression
- Offer to examine ears for chronic serous otitis media and teeth for dental malocclusion, which is common
- Offer to plot head circumference, height and weight on special achondroplasia charts (hydrocephalus can be an early complication), and to grade development

12.4.7 Cushing syndrome

Causes of cushingoid features

- The most common cause is iatrogenic – secondary to exogenous glucocorticoids
- Non-iatrogenic causes can be divided into ACTH dependent and ACTH independent

ACTH dependent

- Cushing's disease (pituitary ACTH-secreting tumour)
- Ectopic ACTH-secreting tumour

ACTH independent

- Adrenal adenoma/carcinoma
- McCune–Albright syndrome

Suggested examination scheme for cushingoid children

General observations

- Growth: offer to plot weight and height, comment on truncal obesity if present, presence of facial plethora and fullness
- Well or unwell
- Look for any sign of disease process potentially requiring steroid treatment – for example, inflammatory bowel disease, allergic bronchopulmonary aspergillosis (ABPA) in children with cystic fibrosis, severe asthma (?evidence of atopy), rheumatic conditions

Arms

- Check blood pressure
- Look for proximal myopathy (both muscle weakness and wasting)
- Thinning of skin

Other

- Moon face
- Buffalo hump
- Evidence of virilisation from adrenal androgens (acne, axillary hair, pubic hair, can also cause clitoromegaly and precocious puberty)

Abdomen

- Striae
- Palpate for masses
- Look for scars (think about adrenal surgery, gastrointestinal disease requiring steroids)

Also …

- Mention you would like to undertake pubertal staging
- Fundoscopy (for pituitary tumour causing optic nerve pressure), visual field examination (for process affecting optic chiasm)
- Suggest full systems examination if systemic disease requiring exogenous steroids is suspected
- Don't use the term Cushing syndrome unless ACTH-secreting tumour or adrenocortical tumour suspected; if unsure refer to findings as 'cushingoid'

12.4.8 Thyroid gland

Suggested structure for examination of the thyroid gland or a 'neck mass'

Instructions to candidate

Examine this child's neck.
Look at this child's neck and tell me what you see.

Initial procedure

- Introduction as previously described
- Ensure that there is a glass of water available

General observations

- Growth
- Well or unwell
- Obvious thyroid signs, eg thin or overweight, eye signs, cold intolerance, etc

Inspection

- From the front with the neck extended, is there a goitre? Is there a visible nodule/mass?
- If a scar is present, listen to the voice for hoarseness
- Ask the child to take a drink; a thyroid swelling should move upwards with swallowing
- Ask the child to stick their tongue out (if it rises during manoeuvre, then it is a thyroglossal cyst)

Palpation

- Palpate the gland while standing behind the child
- Assess size, shape and consistency (single diffusely enlarged thyroid, a single enlarged cyst or a multinodular goitre?)
- Ask to drink while palpating

Further examination

- Examine tongue for high thyroglossal cyst (originates at foramen caecum)
- Palpate for regional lymphadenopathy (eg from thyroid malignancy)
- Percuss sternum for retrosternal extension
- Auscultate mass for bruits

To say at end of neck examination

- I would now proceed to assess the thyroid status.

Suggested structure for examination of thyroid status

Instructions to candidate

Assess this child's thyroid status.
This child has hyperthyroidism; assess her thyroid status.

Hands

- Look for clubbing
- Cool or warm peripheries
- Ask the child to hold out their hands and look for fine tremor: are the hands sweaty?
- Pulse rate and rhythm

Blood pressure

- Wide pulse pressure in hyperthyroidism

Eyes

- Lid oedema
- Exophthalmos/proptosis (examine from above)
- Lid retraction (able to see white of sclera above iris)
- Lid lag (when following examiner's finger in rapid downward gaze, the lids lag behind the iris)
- External ophthalmoplegia
- Conjunctival injection and oedema

Neck examination (as above)

- If not already done

Reflexes

- Slow relaxation in hypothyroidism

Myopathy

- Occasionally get proximal myopathy in hyperthyroidism

Also

- Offer to plot growth (always comment on this as part of thyroid examination)
- Say you would like to stage puberty
- Comment on positive signs, commit yourself to saying whether a patient is clinically euthyroid or hyper/hypothyroid. Remember a patient can have eye signs of Graves' disease but be treated and therefore clinically euthyroid
- Look for other signs of autoimmunity, such as vitiligo, hyperpigmentation (Addison's disease)

- Be prepared to discuss investigations and treatment (see 'Notes on the thyroid examination')

Questions to ask to help assess thyroid status

- Energy levels
- Performance at school
- Heat or cold intolerance, sweatiness
- Constipation, diarrhoea
- Appetite and weight loss or gain
- Muscle weakness

Causes of goitre

- ○ Autoimmune thyroiditis (Hashimoto's disease)
- ○ Graves' disease
- ○ Colloid/simple – normal thyroid function and no antibodies
- ○ Hyperplasia (occurs in iodine deficiency)
- ○ Diffuse nodular non-toxic:
  - Subacute thyroiditis (inflamed gland)
  - Carcinoma
  - Infiltration (Langerhan's cell histiocytosis)

Euthyroid causes of goitre

The most common cause of goitre worldwide is dietary deficiency of iodine, usually seen in developing countries. This results in compensatory hypertrophy. You are very unlikely to see this in the examination in the UK.

More common in the developed world is a child with autoimmune thyroiditis who is euthyroid. Thyroid antibodies are usually present.

Simple or colloid goitres present as diffusely enlarged goitres in peripubertal girls. There are no thyroid antibodies. Multinodular goitres are rare and if seen, malignancy needs to be excluded.

Other causes of neck lumps

Remember other things can be seen on examination of the anterior neck.

It is important to have a list of causes to hand as this is a very common exam scenario:

- ○ Thyroglossal cyst
- ○ Thyroid malignancy
- ○ Haemangioma

- ❍ Sternomastoid tumour
- ❍ Lymphadenopathy
- ❍ Branchial cyst

Causes of chronic cervical lymphadenopathy

- ❍ Reactive lymphadenopathy following oral cavity infection or inflammation: can persist for many months after original inflammatory process has gone
- ❍ The following are rare infectious causes:
  - Atypical TB (mycobacterium)
  - Infectious mononucleosis
  - Cytomegalovirus
  - *Bartonella* (cat scratch disease)
  - Toxoplasmosis
  - AIDS

Thyroglossal cyst

This is a cyst located in the midline of the neck in the route of descent of the thyroid gland from the foramen caecum. They typically rise when the tongue is protruded. It is important to differentiate from an undescended lingual thyroid by radionuclide scanning, as removal of functioning ectopic thyroid tissue may render an individual hypothyroid.

Notes on the thyroid examination

Here is a summary of common thyroid disorders; you may be asked details about these at the end of the examination.

Hyperthyroidism

Causes

- ❍ Graves' disease
- ❍ Hashimoto's disease
- ❍ Toxic nodules
- ❍ Congenital hyperthyroidism – extremely rare, caused by trans-placental thyroid stimulating immunoglobulins from maternal Graves' disease

Presentation is often insidious

- ❍ Sweatiness
- ❍ Heat intolerance
- ❍ Increased appetite with weight loss
- ❍ Poor school performance (poor concentration)
- ❍ Emotional disturbance/agitation

Signs of hyperthyroidism

- Warm sweaty hands
- Fine tremor on holding arms outstretched
- Tachycardia
- Wide pulse pressure and high systolic pressure
- Hyperactive precordium
- Loose stools
- Goitre +/– bruit
- Eye signs – see Box
- Proximal myopathy
- Tall for family height, growth acceleration
- Can cause precocious puberty

Investigations

- TFTs – low TSH, high T4 and sometimes high T3
- Thyroid antibodies – TSH receptor antibodies are positive in Graves' disease. Thyroid peroxidase antibodies are positive in Hashimoto's toxicosis and Graves' disease
- Consider USS neck if goitre present

Treatment

First-line treatment is with carbimazole, and a β-blocker if there are cardiac symptoms.

There are two medical treatment options:

- 'Block and replace': complete suppression of thyroid activity with carbimazole, followed by replacement with thyroxine
- Dose titration: carbimazole doses adjusted to maintain normal T4 levels

Definitive treatment is either by total thyroidectomy or radioactive iodine.

Signs of thyroid eye disease associated with Graves' disease

- Lid oedema
- Exophthalmos/proptosis
- Lid lag (eyelid lags behind eyeball on rapid down gaze)
- Conjunctival injection and oedema
- External ophthalmoplegia

Hypothyroidism

Congenital hypothyroidism

Incidence 1:4000, usually asymptomatic until 6–12 weeks of age due to transplacental passage of maternal thyroid hormone.

Aetiology

- Thyroid dysgenesis (90%) (1/3 thyroid aplasia, 2/3 ectopic thyroid)
- Dyshormonogenesis

This is one of the conditions tested for in the neonatal blood spot screening, and so the signs and symptoms associated are almost never seen in the UK; however it is worth knowing them as this could be a communication skills station and you could get asked 'what if we don't treat'. The most important sequelae are the significant adverse effects on neurocognitive development.

Other clinical features include:

- Hoarse cry
- Constipation
- Lethargy
- Jaundice
- Wide fontanelles
- Macroglossia
- Umbilical hernia
- Developmental delay
- Later … typical facies if untreated

Treatment

This is with lifelong thyroid hormone replacement with thyroxine, monitored with regular thyroid function tests. There is the occasional case of transient hypothyroidism, so a trial off therapy should be carried out in children at the age of 2–3 yrs if they have not required increasing doses of thyroxine as they have grown. If the TSH becomes elevated during this trial off treatment, then thyroxine replacement is restarted.

Acquired hypothyroidism

Causes

- Autoimmune – Hashimoto's thyroiditis
- Iatrogenic – post irradiation or thyroidectomy
- Iodine deficiency (extremely rarely in UK)

Presentation

- Goitre

○ Slowing of growth
○ Pubertal delay/amenorrhoea
○ Lethargy
○ Weight gain
○ Constipation

Signs of hypothyroidism

○ Cold hands
○ Thick skin
○ Bradycardia
○ Slow relaxing tendon reflexes
○ Short for family height
○ increased BMI

Investigations

○ TFTs – T4 low and TSH high. Thyroid peroxidase antibodies (positive in > 90%), thyroglobulin antibodies, TSH blocking antibodies

Treatment

Replacement treatment with thyroxine, monitoring of growth with regular TFTs.

Case 1– Graves' disease

Sarah is a well-looking girl who appears appropriately grown for her age. I would like to plot her height and weight on a growth chart. There is an obvious goitre which moves with swallowing. It is diffusely enlarged, non-tender and firm. There is no lymphadenopathy or retrosternal extension. She has a normal pulse rate and rhythm. Sarah has proptosis. My impression is that she has Graves' disease but she is currently clinically euthyroid. I would like to check her blood pressure and her pubertal staging.

Case 2 – Thyroglossal cyst

Samuel is a well-looking boy who appears appropriately grown for his age. He has an obvious midline swelling of his neck. This is seen to rise on sticking out his tongue but not on swallowing. The mass is smooth and firm. There is no local lymphadenopathy. The most likely diagnosis is a thyroglossal cyst. I would like to check his thyroid status to confirm he is euthyroid.

12.4.9 Puberty

Boys usually start puberty between the ages of 9 and 14 years. The first sign of puberty is attainment of testicular volume of 4 ml. This is followed by the development of pubic and axillary hair. The pubertal growth spurt (peak height velocity) usually occurs at testicular volumes of 12–15 ml.

Girls start puberty earlier than boys; between 8 and 13 years. The first sign is usually breast development. This is followed by pubic hair development, and finally the onset of menstruation.

In both sexes the pubic hair is mostly caused by androgen secretion from the adrenal glands, so-called adrenarche. The sex steroids control the development of other sexual characteristics and are responsible for eventual fusion of the epiphyses and cessation of growth.

The RCPCH short case guidelines state that you will not be asked to examine the genitalia if this would make the candidate uncomfortable. It is therefore unlikely that you will be asked to stage puberty in the examination, but you will need to be familiar with the various puberty stages and be able to describe them (Table 12.5). Remember in boys to think about separate staging of genitalia (penis and scrotum) and axillary hair. In addition, state you would measure testicular volume using an orchidometer; this gives valuable information. In girls, separate staging exists for breast development, axillary hair and pubic hair. These are well covered in most paediatric textbooks and should be revised.

Tanner stages – pubic hair (male and female)	
P1	No pubic hair
P2	Small amount of long downy (fine) hair over labia or base of penis
P3	Adult-type hair – confined to pubis
P4	Adult-type distribution – not extending onto thighs
P5	Hair extends to medial thighs
Tanner stages – axillary hair (male and female)	
A1	No axillary hair
A2	Hair present but not adult amount
A3	Adult amount
Tanner stages – breast development	
B1	Prepubertal
B2	Breast budding
B3	Development of breast mound
B4	Areola projects at an angle to breast mound
B5	Adult configuration – areola and breast in same plane
Tanner stages – male genitalia development	
G1	Prepubertal penis (unstretched length 2.5–6 cm) testes ≤4 ml
G2	Scrotal laxity – no penile enlargement (normally testicular volume 4 ml)
G3	Penile lengthening, scrotal texture more adult
G4	Penile lengthening and broadening (normally testicular volume 10–12 ml), more prominent glans, darkening of scrotum
G5	Adult genitalia (testicular volume 15–25 ml)

Table 12.5: Pubertal staging

Precocious puberty

Remember that changes in growth are closely linked to abnormalities in th pubertal process. True precocious puberty (gonadotropin-dependent precociou puberty) has features of normal puberty, but occurs early. Puberty occurrin, before age of 8 years in girls (ie breast development) and 9 years in boys (i testicular volume ≥ 4 ml) is regarded as abnormally early.

True precocious puberty is more common in girls than in boys and is idiopathi in > 90% of cases. True precocious puberty in boys is uncommon and mucl more likely to be pathological; over half will have some form of intracrania pathology. The psychological problems can be significant. Most will initially b tall and if untreated, will end up with a reduced final height due to prematur fusion of the epiphyses.

Peripheral precocious puberty (or gonadotropin-independent precociou: puberty) can be androgen or oestrogen mediated. It may be caused by congeni tal adrenal hyperplasia, autonomously functioning testes or ovaries glands (e from McCune–Albright syndrome), tumours (eg adrenal virilising tumours- androgens, or juvenile granulose cell tumour–oestrogens). The sequence o development of the signs of puberty is usually abnormal.

The following are **normal variants** of puberty and do not require treatment:

- Premature thelarche: breast tissue development usually in girls under 3 yrs, with no further pubertal progression
- Premature adrenarche: isolated pubic/axillary hair development, no treatment required (but need to consider other causes such as congenital adrenal hyperplasia and adrenal tumour)
- Isolated menarche: no physical signs of puberty with one or more episodes of vaginal bleeding (that cannot be explained by other causes of vaginal bleeding such as vulvovaginitis, foreign body, child sexual abuse, etc.)

An LH-releasing hormone (LHRH) test can distinguish between true (central o gonadotropin-dependent) precocious puberty and peripheral precocious puberty. This involves administering gonadotropin-releasing hormone (GnRH) and measuring the increase in LH and FSH levels. In true puberty there will be a surge in these hormones in response to GnRH administration, whereas in peripheral precocious puberty there will be no rise in LH and FSH.

Delayed puberty

This is common and refers to the absence of signs of puberty by 13 years in girls and 14 years in boys. In the majority it is physiological and is simply secondary to constitutional delay in growth and puberty. A low BMI will also delay the onset or progression of puberty, and so nutritional assessment is an important part of the clinical evaluation of puberty. In girls, Turner syndrome needs to be excluded. In a tall boy with pubertal arrest, Klinefelter syndrome should be considered. If nutrition is optimised with no pubertal development, then these children require further investigation.

Suggested approach to examination of children with delayed puberty

- Look for dysmorphic features (and in particular for signs of Turner or Klinefelter syndrome)
- Make a comment about weight and height, and offer to plot BMI
- Offer to plot the parents' height, and ask the parents about the timing of their own puberty
- If the child looks small and thin, focus the examination on the search for a chronic disease process
- Look for signs of other pituitary hormone problems, such as growth hormone deficiency or hypothyroidism (these children tend to be short and have an increased BMI
- Neurological examination and fundoscopy (looking for papilloedema)
- Examine the visual fields (bitemporal hemianopia can occur in lesions which cause destruction of the pituitary gland such as prolactinoma or craniopharyngioma)
- Make sure that the examiner is aware that you know how to stage puberty (although it is unlikely to be appropriate in this setting)

Investigations to consider for delayed puberty

- Tests directed at underlying chronic disease (eg urea and electrolytes (U&Es), liver function, coeliac antibody screen, inflammatory markers, thyroid function)
- LH, FSH, and oestradiol or testosterone
- Tests of other pituitary hormones (eg prolactin, IGF-1)
- Genetic testing
- In girls, ultrasound scan of pelvis and/or ovaries
- Bone age
- MRI of the pituitary may be required if any concerns are raised by the history or examination

McCune–Albright syndrome

McCune–Albright syndrome consists of abnormalities of endocrine function and skin pigmentation with characteristic fibrous dysplasia of the skeletal system. It is caused by an activating mutation of the GNAS1 and results in permanently 'switching-on' of many G-protein coupled receptors eg TSH receptor, LH receptor, GHRH receptor, MSH receptor and the ACTH receptor. This can cause autonomously functioning endocrine glands and so endocrine hyperfunction, eg hyperthyroidism, Cushing syndrome, peripheral precocious puberty. GNAS1 mutations are somatic, so that different tissues will have different expression of the abnormal protein. This leads to variable manifestations of the condition, with differing combinations of endocrinopathy and bone disease between patients.

Examination findings

- ❍ Abnormal growth: may be tall or short depending on combination of endocrine disturbance/bone disease
- ❍ Large areas of increased pigmentation with ragged 'Coast of Maine' outline
- ❍ May have evidence of precocious puberty
- ❍ Need to examine for signs of hyperthyroidism or cushingoid features
- ❍ Look for evidence of bone disease: deformity from repeated fractures/ evidence of surgery; can have cranial nerve compression
- ❍ May have evidence of osteomalacia/rickets because of increased renal phosphate loss

Congenital adrenal hyperplasia

The most common cause is 21-hydroxylase deficiency, but there are rare forms with varying degrees of adrenal insufficiency. In 21-hydroxylase deficiency there is a block in the pathway that synthesises cortisol and aldosterone, leading to an increase in metabolites which are then converted to androgens. This is driven by adrenocorticotropic hormone (ACTH) secretion in response to inadequate cortisol levels. There is a spectrum of severity, which gives rise to a range of different presentations. In its most severe form, congenital adrenal hyperplasia can present with virilisation, leading to ambiguous genitalia in girls, or a salt-wasting crisis in boys who are a few days old. Milder cases may cause gonadotropin-independent precocious puberty. Diagnosis is usually made by measuring 17-hydroxyprogesterone levels, which remain elevated rather than decreasing several days after birth. Subsequently, genetic testing can be used to look for the common genetic causes of 21-hydroxylase deficiency.

Treatment involves replacing cortisol to suppress ACTH and thereby reduce the drive to produce adrenal androgens. This is monitored by measuring growth (too much cortisol causes weight gain and suppresses growth) and 17-hydroxyprogesterone levels.

Patients may also require salt replacement and mineralocorticoid replacement. This is monitored by measuring blood pressure, salt levels and renin levels (which will be high if there is not adequate replacement). Surgery may be needed to correct ambiguous genitalia. The parents will require pre-conception counselling to prevent future affected girls having ambiguous genitalia (the mother will need to take steroids during the pregnancy). Counselling may be needed if there are body image problems associated with excessive androgenisation in girls.

All patients with congenital adrenal hyperplasia are at risk of adrenal crises if steroids are not increased during times of stress or illness.

12.5 Nephrology

12.5.1 Introduction

Renal cases may be used in the 'other' station or in the abdominal station, eg renal mass palpable on abdominal examination.

The commonest scenarios you are likely to come across are:

- **Child with a renal mass.** The key point is to differentiate the renal mass from a mass due to other causes, eg spleen, faeces; this is covered in Section 9.2. Palpate for a bladder as part of your examination; a large bladder would be due either to bladder outlet obstruction or a neuropathic bladder. Remember a palpable megarectum may appear to be the bladder but will indent.

  If you are sure the mass is renal, **polycystic renal disease** is the most likely cause, although a full differential diagnosis should be known. **Prune belly syndrome** may also be seen (undescended testis, abdominal wall muscle defect and dysplastic urinary system with dilation), with some children having a co-existing congenital heart defect.
- **Child with nephrotic syndrome.** This is one of the commonest renal cases seen in the exam, as such, children are often used for a clinical examination session and a history taking/management planning session on the same day if they are amenable.

  It definitely pays to know nephrotic syndrome well. The physical signs to elicit can be either those of nephrosis or those of **steroid toxicity** and both should be learnt.
- You may see a child with **Chronic Kidney Disease (CKD)** with signs including short stature, nutritional impairment and pallor, and who may have either a peritoneal dialysis catheter or fistula for dialysis.
- Children are also sometimes seen **post renal transplant.** Renal transplantation in the paediatric age group is rare, but such children would be very likely to come to the exam if well and able, and could alternatively attend the history taking and management planning section.

  It is important to look for the side-effects of immunosuppression and to remember that there may be nephrectomy scars if the diseased kidneys have been removed. The transplanted kidney is likely to be in the iliac fossa and is superficial and therefore palpable.
- **Child with loin scars.** It is important to be aware that loin scars are often seen following nephrectomy for poorly functioning dysplastic or non-functioning renal tissue in childhood. The reason for the nephrectomy is the risk of sepsis if there is ongoing reflux to the poorly functioning moiety, and the risk of hypertension long term.

  You may also see a child with Beckwith–Wiedemann syndrome who has had a partial or complete nephrectomy to remove a Wilms' tumour.

Alternatively you may see a child who has had a *laparoscopic* nephrectomy, with port sites instead of the larger loin scar – this is now the more common procedure

Tip +

If you see a renal patient it must be stressed here that measurement of blood pressure is very important. The correct technique is covered in Section 8.2 (cardiovascular station) – however, the underlying cause of hypertension in childhood is often renal.

If you are asked to measure blood pressure and you struggle to do this you might fail the exam!

12.5.2 Renal masses

Case

> *Carly is a thin-looking 15-year-old girl with bilateral and ballotable loin masses. I cannot get above them, they are resonant to percussion and do not move with respiration.*
>
> *I think these masses are renal.*
>
> *She is otherwise well and has a normal blood pressure for her age. I would like to plot her height and weight on a centile chart.*

Supplementary questions

- What is the most likely diagnosis? (Autosomal-dominant polycystic renal disease as bilateral, otherwise well)
- What is the differential diagnosis of a renal mass? (See below) What is the initial investigation of choice? (Renal ultrasound)

Differential diagnosis of a renal mass

- Autosomal-dominant polycystic renal disease
- Hydronephrosis
- Cyst
- Multicystic dysplastic kidney
- Wilms' tumour
- Neuroblastoma
- Compensatory hypertrophy in unilateral renal agenesis
- Renal vein thrombosis
- Storage disorder
- Beckwith–Wiedemann syndrome
- Sepsis/abscess

Notes on polycystic renal disease

Autosomal-dominant polycystic renal disease

The most common of the hereditary kidney diseases. Of adults with chronic renal failure, 8% have autosomal-dominant polycystic renal disease, which is due to mutations in genes PKD1, 2 or 3.

The cysts are in fact grossly dilated nephrons, which progressively enlarge. This leads to compression of the surrounding parenchyma, eventually rendering it ischaemic. Cysts can occur from the prenatal period onward, but the condition commonly presents either with an asymptomatic renal mass in late childhood, or with a mass together with complications in early adulthood. It was formerly called 'adult polycystic kidney disease', but this term is no longer used.

Complications

- Abdominal pain (stretching of cyst leads to pain, but cysts can also haemorrhage or rupture and cause acute pain)
- Haematuria
- Proteinuria (relatively rare)
- Rucurrent urinary tract infections
- Hypertension
- Chronic renal failure
- Intracranial aneurysms in later life
- Heart valve abnormalities later in life, eg aortic regurgitation

Genetic testing is available but not performed routinely; ultrasound is the tool that is currently used for screening first-degree relatives.

Treatment is supportive, eg controlling hypertension with an ACE inhibitor.

Autosomal-recessive polycystic renal disease

This is much less common (1 in 40 000) and is due to mutations in the PKHD1 gene on chromosome 6. Genetic testing is again available in certain circumstances.

The cysts are in the collecting ducts of the nephrons. Can present in the neonatal period with bilateral renal masses and often respiratory distress, due to the pulmonary hypoplasia which developed in utero. However, some cases clinically present in infancy or in childhood.

Congenital hepatic fibrosis is present in all patients, and hepatic cysts are common. Portal hypertension can develop. Systemic hypertension secondary to the renal disease requires aggressive treatment. The prognosis is poor; a number survive several years but inevitably progress to dialysis and renal transplantation.

12.5.3 Nephrotic syndrome

This is a likely case. The child may have few signs and the examiner may give you a lead into the diagnosis (see below).

Instructions to candidate

Examine this 4-year-old boy who has nephrotic syndrome.

Case

John is 4 years old. He looks small for his age; I would like to plot him on a growth chart. On inspection he appears pale and oedematous with a swollen abdomen. He also has bilateral ankle and leg swelling.

He is well perfused peripherally and centrally, and is well hydrated.

His pulse is 96 beats per minute and is of good volume. I cannot measure his JVP because of the oedema. Heart sounds are normal with no added sounds. He is not in respiratory distress and his chest is clear with no clinical signs of pleural effusions. His abdomen is distended. There are no scars or spider naevi. There is no organomegaly. I was able to elicit shifting dullness.

I would also go on to assess his genitalia.

I would like to take his blood pressure, dip his urine and assess him for signs of steroid toxicity.

Supplementary questions

How would you confirm your diagnosis?

I would check a urine dip for protein in the first instance. I would confirm the diagnosis by confirming proteinuria (early morning protein:creatinine ratio > 200) and hypoalbuminaemia (< 25 g/l) in an oedematous child.

If he presented with abdominal pain, how would you assess him?

I would take this sign very seriously. Abdominal pain is a common symptom due to splanchnic vasoconstriction as a consequence of intravascular depletion. The other cause of abdominal pain in children with nephrotic syndrome is peritonitis. I would clinically assess circulating volume and consider sepsis, then treat accordingly. I would consider and treat intravascular depletion if present. Urinary electrolytes (which would show a low sodium/high potassium indicative of sodium depletion) may be helpful.

Notes on nephrotic syndrome

A good knowledge of nephrotic syndrome is required for all parts of the exam, as stressed before. More detailed notes and a thorough management plan can be found in Part I, History Taking and Management Planning. The candidate should also access recent review articles on the subject.

Physical signs of acute nephrotic syndrome

- Oedema
- Pallor
- Tachypnoea and dyspnoea if there is pulmonary congestion
- Pleural effusions and ascites
- Peripheral, including scrotal or vulval, oedema
- Hypertension
- Evidence of poor circulating volume (see Box)

Clinical assessment of the circulating volume

- Blood pressure and pulse (rate and volume)
- Jugular venous pressure (difficult if oedematous or uncooperative young child)
- Core–peripheral temperature difference
- Central capillary refill time

Complications of nephrotic syndrome

- Hypertension
- Acute renal failure/chronic renal failure
- Infection
- Thrombosis
- Malnutrition
- Hyperlipidaemia

Management of nephrotic syndrome

- Use of 5% or 20% human albumin solution for ongoing hypovolaemia or severe symptomatic oedema or ascites (discuss with renal team)
- Steroid therapy – prednisolone 60 mg/m^2 for 4 weeks (or until remission if you are treating a relapse) followed by 40 mg/m^2 on alternate days for a further 4 weeks before stopping

- Treat relapses with further prednisolone and consider cyclophosphamide or ciclosporin for steroid-dependent or steroid-resistant cases

Consider referral for renal biopsy if

- Steroid resistant, ie does not go into remission by 28 days
- Age < 12 months or > 12 years
- Persistent haematuria
- Hypertension not secondary to circulatory problems
- Renal impairment (not improved with correction of circulating volume)
- Low complement

Main histological subtypes of nephrotic syndrome

- Minimal change disease – most common at around 75% cases
- Focal segmental glomerulosclerosis
- Mesangiocapillary glomerulonephritis (also known as membranoproliferative)
- Membranous nephropathy
- Congenital nephrotic syndrome – first 6 months
- Other causes of a nephrotic picture

12.5.4 Steroid toxicity

Instructions to candidate

Examine this boy and tell me what your findings suggest.

Case

Jason is a 12-year-old boy who appears short and obese and I would like to plot him on a centile chart. On general examination he has a moon face and buffalo hump. There is distension of his abdomen with striae and bruising, and he has poor muscle bulk. Jason has hirsutism and hypertrichosis, there is a central line and a nasogastric tube in situ, and a scar on his abdomen in the right groin with a palpable mass underneath it.

These findings suggest that Jason has iatrogenic Cushing syndrome secondary to steroid-induced immunosuppression following a renal transplant. Hypertrichosis suggests he is also taking ciclosporin.

I would like to measure his blood pressure and dipstick his urine for glucose.

Notes on steroid-induced toxicity

The clinical features of steroid toxicity ('cushingoid' child)

General

○ Growth retardation with truncal obesity
○ Oedema
○ Hirsutism

Face and eyes

○ 'Moon face'
○ Acne
○ Cataracts

Trunk

○ Buffalo hump (centralised fat distribution)
○ Abdominal striae and bruising

Limbs

○ Poor muscle bulk
○ Proximal myopathy

Tip +

Other side-effects of steroids include glycosuria, osteopenia and psychosis

Aetiology

There are various scenarios that can lead to steroid-induced toxicity that you may meet in the exam. Examples include:

- Asthma
- Juvenile chronic arthritis
- Cystic fibrosis
- Dermatomyositis
- Crohn's disease
- Post renal/cardiac transplant (look for relevant scars)
- Nephrotic syndrome

12.5.5 Chronic kidney disease

Instructions to candidate

Examine this 13-year-old boy and tell me your positive findings.

Case

Callum is a slim boy of relatively short stature. I would like to plot his height and weight on a centile chart. He looks slightly pale.

On inspection of his hands, he has multiple scars from cannula insertions.

On examination of his abdomen, he has a peritoneal dialysis catheter that is capped off. He has no scars on his abdomen or in either loin.

... I think he has chronic kidney disease and is having peritoneal dialysis ...

I would like to check his blood pressure and a urine dip ...

Notes on chronic kidney disease

This can be defined as a persisting bilateral renal injury or a GFR less than 90 ml/min/1.73 m^2 for over 3 months. By the time this is diagnosed there will have been a variable degree of renal parenchymal damage, which is irreversible.

The CKD is staged 1–5 according to GFR. The extremes are:

- Stage 1 = GFR > 90 ml/min/1.73 m^2, but evidence of bilateral renal injury
- Stage 5 = GFR < 15 ml/min/1.73 m^2, which is termed renal failure

Some of the commonest diseases causing CKD are:

- Congenital abnormalities of kidneys or urinary tract
- Renal cystic disease
- Nephrotic syndrome
- Glomerulonephritis

Puberty is often delayed, and a proportion of children will go on to need renal replacement therapy (RRT). The main management principles are:

- Preventing progression of proteinuria (with ACE inhibitor) and monitoring early morning urine protein:creatinine ratio, serum electrolytes and creatinine.
- Controlling hypertension – salt reduction, anti-hypertensives such as ACE inhibitor.

- Safe management of intercurrent illnesses – avoiding dehydration and not using nephrotoxic drugs, eg ibuprofen. Some drug doses will need to be lowered if their use is critical (eg aciclovir).
- Fluid and electrolyte management, including dietary intake, tailored to the child's particular condition. If polyuric from a tubular defect they will need supplementation of sodium and bicarbonate plus relatively larger volumes of water intake. However if a child has a glomerular defect they can retain salt and water, and need to lower sodium intake. A dietician is very important here.
- Prevention of metabolic bone disease – reduction of phosphate intake, Vitamin D supplementation.
- Monitoring and treating growth and nutritional problems. Poor growth is often seen in CKD and can be exacerbated by poor oral intake,which can be due to anorexia, nausea and vomiting. Regular dietetic reviews and calorie supplementation/enteral feeds may be needed. Some children are best managed with a gastrostomy.
  Those with particularly poor growth despite optimal management of their CKD may benefit from growth hormone.
- Correcting anaemia, which can be very debilitating, by using erythropoietin and iron supplementation. Reduces need for multiple transfusions and the risks these entail.
- Immunisations as per primary schedule, plus annual influenza vaccine.
  If being worked up for transplantation, need live vaccines in run-in to transplant as they cannot be given afterwards.

12.5.6 Post-renal transplant

Instructions to candidate

Examine this boy's abdomen.

Case

Stanley ia a 6-year-old boy who looks short for his age – I would like to plot his height and weight on a centile chart.

There is a scar in his right iliac fossa, and scars in both loins on inspection at the back. He has a soft, non-tender abdomen with a palpable mass in the right iliac fossa.

I can get above and below it. It is dull to percussion.

I think this is a transplanted kidney.

I would like to check his blood pressure and dip his urine for protein.

Supplementary questions

What complications can occur in these children?

- Infections more likely, secondary to immunosuppressive therapy
- Rejection of transplanted kidney
- Dehydration with intercurrent illness – risk of acute kidney injury, so lower threshold for iv fluids and consider iv steroids to avoid adrenal crisis nephrotoxicity
- Obstruction (ultrasound of transplanted kidney, check renal pelvis measurements pre and post micturition)

Other points to remember in management are:

- Live attenuated vaccines, including nasal influenza (not the subcutaneous injection) and oral polio are contraindicated.
- Prophylaxis with co-trimoxazole (*Pneumocystis jiroveci*) and valganciclovir (if high risk for CMV) in the few months immediately post transplant
- Varicella zoster exposure. If non-immune, vaccinated before transplant and immunity regularly checked. If contact post transplant and non-immune, varicella zoster immunoglobulin given. If chickenpox, give iv aciclovir.
- Avoidance of drug-induced nephrotoxicity, eg NSAIDS.
- Clinic reviews – weight, BP, JVP, laboratory testing, including regular monitoring of creatinine vs 'baseline' creatinine.

12.5.7 Beckwith–Wiedemann syndrome

Instructions to candidate

Examine this girl who presented in the neonatal period with profound hypoglycaemia.

Case

Zoe looks large for her age – she is 2 years old. I would like to plot her height and weight on a centile chart. On inspection of her face she has transverse ear lobe creases and macroglossia.

On abdominal examination I notice she has a scar adjacent to her umbilicus, and a nephrectomy scar in the right loin. There is no organomegaly.

There is no obvious hemihypertrophy on examination of her limbs.

*The diagnosis is **Beckwith–Wiedemann** syndrome and her scars are from surgery on her exomphalos in the early neonatal period, and from a partial or complete nephrectomy which may have been required due to a Wilms' tumour.*

Notes on Beckwith–Wiedemann syndrome

This is a rare fetal overgrowth syndrome (around 1 in 14 000 live births) associated with gene locus 11p15.5. Most cases are sporadic, although there have been clusters of cases within families with an autosomally dominant distribution.

Clinically, there are major and minor features. Diagnosis is based on:

1. Three major features, or
2. Two major **plus** three or more minor features

Major clinical features

- Pre- and/or postnatal overgrowth (> 90th centile)
- Macroglossia
- Abdominal wall defects

Minor clinical features

- Characteristic ear signs – ear lobe creases or posterior helical pits
- Facial naevus flammus ('stork mark')
- Hypoglycaemia
- Organomegaly
- Hemihypertrophy

Natural history of disease

Infants are more likely to be delivered prematurely, 35% before 35 weeks. **Exomphalos** occurs in 50% of cases and **hypoglycaemia** is common, which is usually mild and transient, requiring frequent feeding only. However, some babies have severe hypoglycaemia, requiring diazoxide. Deaths from Beckwith–Wiedemann syndrome mainly occur in infancy and are usually caused by problems related to prematurity or associated congenital cardiac disease (< 10%).

During **childhood**, the dysmorphic features become less apparent, although the macroglossia may cause feeding problems, problems with speech (requiring speech therapy) and occasionally with obstructive apnoea. Surgical tongue reduction may be required in severe cases.

Overgrowth is most marked in the first few years and is associated with an advanced bone age. It tends to slow down in late childhood, and most adults are < 97th centile. **Hemihypertrophy** occurs in 25% of cases, and visceromegaly is common.

Neoplasias occur in 5–10%, most commonly with Wilms' tumour followed by adrenocortical carcinoma, hepatoblastoma and neuroblastoma. Children with hemihypertrophy are most at risk and those who are going to develop a tumour usually do so in the first decade of life.

Some children have severe learning difficulties, which are thought mainly to be from neural damage secondary to neonatal hypoglycaemia rather than congenital central nervous system malformations.

Screening

Alpha-fetoprotein levels (hepatoblastoma marker) are monitored until 4 years of age.

Most centres advocate regular ultrasound examination until at least 8 years old. Wilms' tumours can present rapidly in younger children and mean size at presentation is reduced significantly if the child has 3- to 4-monthly screening. Small tumours can be removed with preservation of renal function.

The other risk factors for Wilms' tumour are important and should be known, as the association may arise as a supplementary question in a short case:

- Beckwith–Wiedemann syndrome
- Hemihypertrophy
- Aniridia
- DRASH syndrome (ambiguous genitalia nephropathy, Wilms' tumour)

Prognosis

Variable. Severely affected children are at risk of death or major morbidity eg from hypoglycaemia, tumours.

By adolescence, the majority of surviving children can lead a normal life as long as there are no sequelae of neonatal hypoglycaemia.

13

CHILD DEVELOPMENT

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13.1 Introduction

The development station can be a very challenging section of the exam. It is often the station candidates are most anxious about, which is understandable as it is the examination most will perform least frequently in their day-to-day working life. In fact, of all the stations it is the most predictable. You will be asked to examine a child between the ages of 0 to 5 years. Usually you will be asked to examine one or two specific areas of development as there simply isn't time to assess all areas of development. You may however just be asked to 'assess this child's development'. You then need to make a judgement as to where to start – usually the most appropriate starting point is to look at gross motor development. You must also be prepared to comment on other areas of development.

It is absolutely essential you are fluent in the developmental milestones (see Tables 13.1–8). As with all the stations, it is about getting as much practice as you can in carrying out developmental assessments. Team up with a colleague and start with as many developmentally normal children as you can. Be observed and timed. Most candidates find that they are given a child to examine who is delayed in at least one area, though you should also be prepared to be asked to examine a child who has no developmental delay whatsoever.

Be familiar with the Anchor Statements provided by the RCPCH while you are preparing. You are being assessed on your ability to give clear instructions to the child, to ask about parental concerns and to use history taking where appropriate, but this should 'not replace the examination'. You are also being assessed on your ability to give a clear summary of your relevant findings, and to suggest the next step in examination or investigation/referral.

The following are some practical points about how your style and approach can reassure the examiner and the parents that you know what you are doing, and help you to maximise your performance in this station:

○ Structured systematic approach (start at just below estimated age and work up until just above expectations).

○ Ask questions – but these need to be structured and focused, ie pertinent to the specific case. When preparing by practising your examination technique, try to rank your questions in order of importance and see if your colleagues agree with your choice. The most important questions relate to your ability to effectively conduct an examination; if giving instructions to a child you need to know 'can they hear and understand me?' Other relevant questions relate to details of their development that you can't assess there and then (eg toileting/climbing stairs), or checking their medical/developmental history to get a clearer understanding of their difficulties (were they premature? age of first words/walking?, etc).

- Start by getting down to the child's level and begin playing with the child and observing/commenting on what they are doing. Opportunistic observations and comments are key – if you have already seen the child crawling as you entered the room there is no point in wasting time during your examination getting them to demonstrate it again. Although remember to tell the examiner you observed it when you present your summary of the case.
- When introducing new activities, clear the area. If you want them to build a tower you don't want them distracted by pens and paper on the same table. Comment on what the child does when not being directed, eg if introducing bricks, comment on what they do initially before trying to get them to build a tower. Similarly if producing pen and paper, watch what they do and comment before directing them to draw a circle.
- Start with easy tasks and progress to more difficult ones. For example, if assessing fine motor skills, start with using bricks, next move on to using pen and paper, then threading beads, and finally using scissors. Demonstrate what you expect them to do (makes instructions easier for them to understand).
- Give clear instructions to the child. If you say to a small child 'would you like to?', and they say 'no', your structured approach will fall apart. You need to ask a child to do something in a way that sounds polite, natural and interesting for them, and does not provide them with an option to refuse. As in clinical practice, with very young children, physical examinations are done with the parent's not the child's consent. Having said that, if the child is uncomfortable or uncooperative the examination will be hampered, so skilled persuasion and distraction should be practised. Above all, be nice to the child and their parent!
- Find something the child can do and something they can't do, so that you have a minimum and a maximum age. The aim is to assess developmental age to within 2–3 months up to 2 years, and within 4–6 months for 2- to 5-year-olds.
- Watch your choice of language. The examiners will be very mindful of how you are coming across to the child's parents. Practise using phrases such as 'today Jonny did not demonstrate…' rather than 'he cannot…'

Conditions commonly seen in the exam

Autism spectrum disorder
Down syndrome
Non-mobile toddler
Global developmental delay
Hypotonia
Hemiplegia/diplegia
Developmentally normal children

13.2 Clinical examination

Suggested approach to developmental examination – infant

General observations

- Well or unwell?
- Growth?
- Any obvious dysmorphic features?
- Any equipment that gives hint of delay, for example nasogastric tube, special chair

The area of development you then concentrate on will be led by the examiners' instructions and the position and co-operation of the infant. In general, you should always start by observing and commenting on as much as possible.

Gross motor examination (posture and movement)

Remember muscle control progresses from the head down. Suggested system for a baby could be:

- Lie the child supine and assess their general tone/limb positions and movements. Can they roll? (You could comment on vision at this stage)
- Gently pull on the hands or arms until the baby is in sitting position (for 3 to 6-month-old children): look for head lag, assess straightness of spine, upper limb and truncal tone, can they sit unsupported?
- Hold the child in ventral suspension: comment on truncal tone, head lifting, position of limbs
- Place the child prone and observe their ability to lift their head/torso/roll over or crawl away
- Finish by assessing the presence of primitive reflexes (see Box)

Mobile infants (rolling/pulling to stand/cruising) will not co-operate with this approach and are best assessed by just observing what they do.

Primitive reflexes

- ○ **Asymmetric tonic reflex:** present from birth to 4–6 months. Abnormal if fixed in one position, to one side or if persists at 6 months
- ○ **Grasp:** present from birth to 4 months. Child develops the ability to grasp voluntarily as the reflex disappears
- ○ **Moro:** present from birth to 4 months. Pathological if present beyond this
- ○ **Stepping:** present from birth to 6–8 weeks of age; reappears when child starts walking at 10–15 months (voluntarily)
- ○ **Downward parachute:** develops between 6–12 months of age and persists. Abnormal if not present by 12 months

Fine motor (vision and manipulation)

- In a baby, try to ascertain whether the baby can fix and follow. Remember you must be silent when you are doing this as babies will turn their heads to sound and this can be misleading
- Use a toy or interesting object and watch how the baby grasps and explores the object
- Look for passing from one hand to another and mouthing. Ensure that both hands can grasp equally well. In older infants you could comment on their ability to let go of a toy, and object permanence
- Use something small (eg tiny piece of paper) to assess pincer grip
- Comment on pointing

Speech

- Comment on any vocalisation you hear

Hearing

- In practice you would ask the parents whether they have any concerns about a baby's hearing; the examiner may be happy for you to do this. Make sure you have commented on what you can see and hear first. If the baby clearly startles to a loud noise then comment on it!
- There are other key questions that provide evidence of hearing development; for example, recognising their own name/mamma and dadda sounds – are these used in context or just as babble? Be familiar with these so you can ask them if you are given the opportunity

Social

- Watch the infant's interactions with you and with their parent; are they smiling?
- Look for the presence or absence of stranger awareness
- Make sure the child and parent are comfortable with you before you leap in with waving/clapping/playing peek-a-boo, etc

And finally

- Have some questions prepared in case the examiner lets you ask them. If allowed, then concentrate on social and hearing questions as these are difficult to assess fully in an exam environment
- Present your findings in a structured, ordered manner – this will help you to remember your findings as well as being easier for the examiner to take in
- Think about appropriate investigations or referral if any abnormality is found.

Case – normal 6-month-old boy

Jake is a 6-month-old boy who has normal gross motor and fine motor development.

Jake demonstrated rolling from front to back. When pulled to a sitting position he has a straight back and good head control. He sits briefly unsupported. He weight-bears when held in a standing position. He has no Moro reflex. He did not demonstrate that he could crawl or pull to stand, and his mother reports that he has not done either of these yet.

He reached out for a brick and demonstrated transferring it from hand to hand. He also demonstrated mouthing.

I would like to ask his mother:

- *if she has any concerns about his hearing, and whether she feels he responds to his name*
- *if he is babbling*

as he did not demonstrate these during the assessment today.

Milestones – infant

Note – age ranges are to be used as a guide. You will notice different texts have slightly different quoted ranges. Development occurs in a logical sequence and this is more important than the age range for each milestone. You should be clear about the ordered progression of development; you should also know specific ages at which, if certain key milestones have not been met, further investigation or input is required.

Infant milestones are listed in Tables 13.1–13.4.

Time	Milestones
6 weeks	• Turns head to one side when supine • Asymmetric tonic reflex present • Jerky arm movements • Some head lag/incomplete head control • In ventral suspension, head held in same plane as body momentarily, hips extended • Chin intermittently lifted off couch when prone • Moro present
3 months	• No head lag when pulled to sit • Strong kick • Bears weight on forearms and partly lifts torso off floor when prone; head to 45° and looking around • In ventral suspension: head held above rest of body, looking around
6 months	• Spontaneously elevates head when supine • Lifts head when about to be pulled up to sitting position • Sits with straight back – if not unsupported, then may show righting reflexes • Sits unsupported (5–7 months) • Rolling (2–6 months) • Lifts entire torso off floor when prone, bearing weight on hands (arms extended); head to 90° • Bears weight on legs, when held in standing position (3–7 months) • Can reach for toy
9 months	• Sits unsupported • Crawling (6–9 months) • Pulls to stand (6–10 months)
12 months	• Gets to sitting from lying position (6–11 months) • Cruising (7–13 months) • Stands independently (11–14 months) • Walks independently (10–15 months)

Table 13.1: Infant milestones: Gross Motor

Time	Milestone
Birth to 9 months	• Stares (at birth) • Fixes and follows (6 weeks) • Reaches to grasp (palmar grasp) (3–6 months) • Mouthing (from 4 months) • Grasps, brings to midline, then mouths (6 months) • Transfers hand to hand (6 months) • Fixes on small objects (5–8 months) • Follows fallen toys (4½–8 months) (object permanence)
9–12 months	• Throws (9–15 months) • Pincer grasp (9–14 months) • Bangs bricks together (7–13 months) • Points with index finger (12 months) • Scribbles (12–24 months)

Table 13.2: Infant milestones: Fine Motor and Vision

Time	Milestone
6 weeks	• Comment on noises/vocalisation • Startled by sudden noise/quietens to human voice
3–6 months	• Turns to sound
6 months	• Turns to mother's voice/laughs/babbles
From 4–9 months	• Imitates speech sounds
6–9 months	• Mamma/dadda sounds (used non-specifically)/combining syllables
12 months	• Responds to own name/understands simple commands
7–13 months	• Uses mamma/dadda appropriately
11–15 months	• First word (not mamma or dadda)

Table 13.3: Infant milestones: Hearing and Speech

Time	Milestone
6 weeks	• Smiles
6 weeks	• Looks at faces
6–8 months	• Stranger awareness
5–10 months	• Plays peek-a-boo
8–13 months	• Waving
10–15 months	• Indicates wants (not by crying)
9–12 months	• Casting
9–12 months	• Clapping

Table 13.4: Infant milestones: Social

Suggested approach to developmental examination – older child

Instructions to candidate

Assess this child's development.

This child has Down syndrome. Assess his fine motor development.

Assess this child's speech and language development by talking to her and her mother.

Assess this 3-year-old's gross motor development.

Initial procedures

- Introduce yourself to the parents and child.
- If not already told, ask whether you can ask the parent some questions.

Some key questions

- Do you speak English at home? (If seems relevant!)
- Do you have any concerns about your child's vision or hearing? (Important if you are going to be asking a child to copy or follow instructions)
- Has your child got any pain anywhere?
- Was your child born at term or were they premature?

Note: if there is some time after the clinical assessment, or if the child is completely unco-operative, you may get an opportunity to ask a developmental history, so have some questions in mind.

General observations

Where relevant, make a comment on the following:
- Growth
- Well or unwell

If there are no obvious things to comment on in the following areas, save time by not mentioning them!
- Equipment (wheelchairs, nasogastric tubes, hearing aids, etc)
- Obvious dysmorphic features

Gross motor examination (posture and movement)

What is the child doing now?

Observe and comment in less co-operative or less directable children. Think about:
- Gait
- Running
- Climbing

For older children think about asking them to:

- Jump up high
- Stand on one foot
- Kick a ball
- Bounce and catch a ball
- Ride a tricycle
- Climb stairs if stairs available

There will be equipment provided to assess gross motor function in older children, so look for tricycles and balls when you get into the room and don't forget to use them if they are appropriate. Some of these activities may be best left towards the end of the examination, for example once you have encouraged them to run about and kick a ball they may not want to stop and do something that requires more focus.

Remember to join in and look like you are having fun, no matter how stressed you are. Be enthusiastic and congratulate them!

If you get a chance to ask the parents a question then you could ask, 'how does your child go up and down stairs?' You may need to follow this up with specific options. Do they walk upstairs one foot per step? This will give you a clear idea of gross motor functions.

Fine motor and vision

This may be a good place to start your examination, particularly if the child is focused on a 'sitting down' activity when you arrive in the room.

Always start by watching and commenting before directing the child towards more specific activities. A suggested sequence is as follows:

1. Picture book:
   - This is better suited for younger toddlers
   - Look for pointing, recognising objects and turning pages
   - Comment on vocabulary and speech, as this can often be assessed using the pictures in the book
2. Drawing (if pen and paper handy) – 'Are you good at drawing? Let's draw something together':
   - Note whether the child can imitate (child watches as you draw a shape) or copy (paper covered and the child cannot see what you are drawing until you are finished). As a general rule, imitating comes approximately 2 months before copying. Make sure you use the right terminology when reporting findings.
   - If you are in the middle of a full developmental assessment, you may well be able to get all the information you need to comment on developmental stage with this activity alone. You could then quickly move on to other areas. If you have only been asked to assess fine motor and vision, then spend some more time on this activity.
3. Building blocks – 'Let's do some building – can you do this?':
   - Remember the difference between copying and imitating as described above.

4. Other activities – you may have been supplied with equipment to assess:
    - Threading beads
    - Using scissors
    - Matching colours

Hearing and speech

- Pointing to body parts
- Ask about name, age, boy or girl, counting, birthday
- Comment on vocabulary and how easy the child is to understand

Social

This aspect of development can be difficult to assess in the exam setting as it is all to do with self-care/independence:

- Dressing
- Toileting
- Eating and drinking

Communicating

For older children, ask about activities and friends at school or preschool.

Finally

Structure your presentation of the developmental assessment into the different areas of development. Be systematic.

First, comment on 'developmental age' for each area of development you have assessed:

In the area of gross motor skills, he has a developmental age of …, because he can …, but today he did not demonstrate … In the area of fine motor and vision …'

Next, comment on whether you feel this is appropriate to the child's chronological age, or whether you feel it is delayed. Be aware of the limitations, as well as the significance of your findings.

Suggest a next examination based on what you have noted, eg neurological exam of lower limbs/spine, assessment of child's vision, etc.

If the child has delayed development, be prepared to discuss investigations/referral – although not in detail.

Milestones – older child

Age or activity	Milestone
18 months	• Running, bending over • Walks carrying toy • Pushes/pulls • Climbs onto a chair
2 years	• Stands on tip toe • Squats and stands up • Jumps up (in place)
3 years	• Walks on tip toes • Briefly stands on one foot (for 1 s) • Jumps with both feet together (30 months) • Runs fast • Rides a tricycle – pushing pedals (there may be a tricycle in the room!)
4 years	• Hops • Stands on one foot for 3–5 s • Ask mother if child can climb a ladder
Gait	• Broad based – 15 months • 18 months – purposeful and confident gait
Ball skills	• Kicks ball (15–24 months) • Can run to kick a ball (4 years) • Bounces and catches ball (5 years)
Stairs	• Crawls upstairs – from 15 months • With help (holding onto adults hand) 21 months • Climbs with two feet per step holding onto rail – 2 years • Climbs with one foot per step – 3 years • Descends with two feet per step – 3 years • Up and down stairs in normal 'adult' fashion/running upstairs – 4 years

Table 13.5: Milestones for the older child: Motor

Time	Task
10–18 months	• Grasp – delicate pincer grasp
12–18 months	• Enjoys simple picture books
18 months	• Turning pages
2–2½ years	• Removes wrappers from sweets
3 years	• Cutting paper
3 years	• Threads large beads onto shoelace
3 years	• Matches two to three primary colours
	Drawing:
12–24 months	• Horizontal scribble
2–2½ years	• Circular scribble
2½ years	• Imitates vertical line
2½–3 years	• Copies circle
4 years	• Copies cross/square
4 years	• Draws a recognisable house
5 years	• Copies triangle
3 years	• Draws a person with a head
4 years	• Draws with head plus three other parts (two arms = one part)
	Building tower:
15 months	• Two bricks
16–24 months	• Three to four bricks
2 years	• Seven bricks
3 years	• Nine bricks
4 years	• Imitates bridge of three bricks
5 years	• Copies three steps from six bricks

Table 13.6: Milestones for the older child: Fine Motor (vision and manipulation)

Time (years)	Milestone
1–2	At least three words (12–18 months)
	Often constantly babbling (18 months)
	Naming body parts– 'can you point to your ...' (18 months)
	Pointing to pictures (18–24 months)
2	Plurals (find two of something and say 'what are these?')
	Gives name
	Large vocabulary (definitely greater than 20 words)
	Puts two words together
	Can say about 50 words
3	Prepositions? ('Where is the ...?')
	Are you a boy or a girl?
	How old are you?
	What colour is the ...? (can identify at least two colours)
	Uses sentences of four words
	Speech can be understood by strangers
	Can follow three-step instructions
4	'Let's do some counting ...' (up to 10 = 4 years)
	Speech is grammatical
	Gives first and last name
	Can name shapes
	Holds a conversation
	Can tell a story in the past tense and in the future tense
	Knows more than two personal pronouns (I, you, he, she, etc)
	Knows several nursery rhymes
5	Fluent and clear speech
	Knows birthday

Table 13.7: Milestones for the older child: Hearing and Speech

Task	Milestone and normal time of achievement
Dressing	• Tries to help with dressing (eg arms into coat) (12–15 months) • Takes off shoes and socks (13–20 months) • Can get dressed with a little help/supervision (2½–3½ years) • Dresses without help (3½–5½ years)
Toileting	• Indicates toilet needs (from 18 months) • Dry during the day (ie out of nappies) (2½–3 years) • Uses toilet alone/washes and dries hands (3 years)
Eating/drinking	• Uses spoon (makes a mess, but can get food to mouth) (14–18 months) • Uses spoon skilfully (18 months) • Drinks from cup (10–16 months) • Eats with spoon and fork (2½ years) • Eats with knife and fork (4 years)
Play/behaviour	• Explores environment (13–20 months) • Tantrums when frustrated (2 years) • Imitates adult activities (2 years) • Plays alone (2–2½ years) • Likes to help adults (3 years) • Engages in imaginative play (3 years) • Shares toys (4 years) • Understands taking turns (4 years) • Dramatic make-believe (4 years) • Chooses own friends (5 years)

Table 13.8: Milestones for the older child: Social

Classic 'warning signs' of developmental delay

Fine motor/vision

- Fixed squint at any age or the development of a new intermittent squint
- Hand preference before 18 months (usually not established until third year)
- Not staring/no fixing or following at 6 weeks
- Immature grip at 18 months
- Still casting at 18 months

Speech/hearing

- Failure to respond to sound at any age
- Not babbling at 12 months
- No single words at 16 months
- Inability to understand simple commands at 18 months
- Unable to string two words together at 2 years
- Unable to understand speech at 2½ years
- Poor articulation, making speech difficult to understand at 4 years
- Any loss of language (regression)

Always remember to mention audiology referral if you notice delay in this area of development (and be aware of how hearing is tested in different age groups, see Section 13.4).

Gross motor

- Asymmetrical neonatal reflexes
- Poor head control at 4 months
- Persistence of primitive reflexes (see page 454 'Primitive reflexes')
- Not sitting unsupported at 9 months
- Not pulling to stand at 12 months

Social/behavioural

- Failure to smile: average age is 5 weeks, should be concerned if not smiling at 6 weeks

Notes on developmental delay

Investigations for developmental delay

You may be asked what investigations you would request following your developmental assessment. Investigating for developmental delay varies between centres and paediatricians. You should tailor your requests based on each individual case, however below is a list of common first-line investigations:

- Chromosomal analysis by array comparative genomic hybridisation (array CGH) – this has the highest yield in developmental delay with no clear aetiology
- Fragile X analysis – this is often difficult to diagnosis clinically as dysmorphic features may be subtle
- U+Es and calcium – hyercalcaemia in DiGeorge and Williams syndrome
- Lead and biotinidase – lead toxicity is treatable and reversible, as is biotinidase deficiency
- Creatine kinase – Duchenne muscular dystrophy
- Thyroid function tests – congenital hypothyroidism is screened for as part of the neonatal screening; however it is worth confirming this is normal as it is a treatable condition that causes significant delay if not treated
- Ophthalmology and audiology assessment

Second-line investigations to consider:

- Metabolic screen – this has a low yield if there are no other features such as developmental regression, organomegaly, coarse facies, a family history, consanguinity
- Neuroimaging – MRI brain if macro/microcephaly, focal neurology, seizures
- EEG – if history of seizures or concerned about a neurodegenerative disorder

Multidisciplinary assessment

Paediatricians are seldom the only professionals to assess a child where there is concern about developmental delay. Remember the other key professionals involved.

Key professionals involved in child assessment

- Paediatrician
- Physiotherapist
- Occupational therapist
- Speech and language therapist
- Portage worker
- Educational psychologist (if a child is likely to have special educational needs or benefit from specialised school or nursery placement)
- Audiology
- Ophthalmology/orthoptics
- Orthotics
- Orthopaedic surgeon
- Neurologist
- Geneticist
- GP
- Health visitor
- Nursery/school teacher

13.3 Visual assessment

Chapter 11, on neurology, contains a description of how to examine the eyes. The following are some additional notes on the assessment of visual acuity. If you have not already had the opportunity try and sit in for a session with an orthoptist and observe them carrying out assessments.

Assessing visual acuity

In the neonate or young infant, examining the eyes for cataracts and the presence of the red reflex should be part of this assessment:

- **12 weeks** The baby should be able to fix and follow an object an arm's length away 180° in the horizontal and vertical planes. (At 6 weeks they can fix and follow to 90°.)
- **10 months** The baby can pick up small objects such as a tiny bit of screwed up paper or a raisin.
- **2–3 years** In the exam setting, use picture books or cards held close up and then further away and see if they can identify simple objects. This is done in a formal manner in the multidisciplinary assessment using standardised picture cards with images of different sizes that are held at set distances from the child and is usually carried out by an orthoptist.
- **4 years** At this age children can often identify letters and a Snellen chart can be used. If there is developmental delay, then some of the previous ways of assessing visual acuity need to be used.

Ideally each eye should be tested separately. In practice this is often not tolerated before the age of 2 to 3 years. To do this the parent's hand can be used to cover one eye at a time, show you are making a good attempt to do this, but if the child becomes distressed, then comment that you 'would like to test each eye separately, however as the child is becoming upset, you will move on.'

13.4 Assessment of hearing

In the lead up to the exam, try and attend an audiology clinic to learn about the assessment of hearing. You may be asked to assess a child's hearing, however it is more likely you may be asked how this is done:

- 0.5–1 newborn/1000 live births have permanent, moderate to severe, bilateral sensorineural hearing loss. In addition children become deaf throughout childhood for various reasons, and the estimated prevalence of permanent, bilateral hearing loss of moderate to severe degree increases to 1.5–2/1000 children under the age of 6 years
- Until children reach the developmental age of 3–4 years, they cannot be expected to respond to a pure tone audiogram, so different methods of assessment must be employed
- You should not be asked to perform a hearing test in the exam as you will soon learn that examination rooms (which contain a lot of children) have a background noise level far above that acceptable for a hearing test. You may be asked to talk the examiner through one of the general principles
- There are two types of hearing tests: those used for the diagnosis of hearing loss, and those used as screening procedures
- A good history from the parents is the first step in a hearing assessment – parental suspicion of hearing loss, family history, risk factors, failed screening test, symptoms of middle ear disease or behavioural problems

Screening for hearing loss

The need for early detection of hearing defects is clear and screening for hearing loss is carried out in all newborn babies in the UK (see www.hearing.screening.nhs.uk).

After the newborn screen, the next routine test of hearing is at school entry. Children undergo a hearing test in the form of a simplified PTA (pure tone audiogram) at a fixed intensity, usually 20–25 dB. Any children failing the assessment are referred to an audiology clinic.

Evoked otoacoustic emissions

- Method used by the universal newborn hearing screening programme in UK
- Sound stimulus produces an acoustic emission from the cochlea that can be detected by a small microphone placed within the ear canal, analysed and displayed; the child needs to be calm and still
- These detectable emissions are only present if the cochlea is functioning and so this test detects most causes of deafness

Distraction testing

By 6–9 months, children have learnt to control their sitting balance and head posture and can turn to sound either above or behind them. They have not yet learnt 'permanence of objects' and so when distracted, do not remember the tester behind their backs.

- Explain the test to the mother/carer
- Position the baby carefully
- The distracter sits in front of the baby and gains their attention with toys
- This activity is then suddenly stopped; the baby waits for the reintroduction of play and the tester, who is standing behind the baby and out of their visual field, presents a sound stimulus
- When the baby turns to look for the sound source they are rewarded with praise. If they do not respond, the tester raises the intensity of the sound stimulus until the baby responds
- Standardised sounds are presented to each side

Pitfalls: the baby can become too interested in the examiner or the baby responds to visual stimulus after play has ceased and becomes used to the test.

Co-operative tests

Distraction testing becomes more difficult after 15 months as the child develops object permanence and becomes more engrossed in play (they are unlikely to turn to even loud noises in the distraction test). From this stage onwards it is usually possible to use co-operative tests, although the age range at which children will co-operate is wide. Here you use either the child's ability to discriminate speech or tasks that demand a response to certain sounds. One of each should usually be performed. It is difficult to assess each ear separately, but to a certain extent this is academic as the immediate aim is to assess functional hearing and the ability to develop language.

Speech discrimination tests

- The McCormick toy test is the most widely used for children 2½ years or more. Here a set of toys is placed in front of the child. Generally toys are paired, with each pair having similar sounding names, eg key and tree, cup and duck, horse and house, spoon and shoe, etc. Having involved the child in play with these toys, the examiner covers their mouth (to stop the child lip reading), and lowers the level of their voice intensity and asks the child to identify one of the objects on the table
- The voice should be at a level of 30–40 dB

Performance tests

Used for children between 2 and 4 years' developmental age. In these tests the child is asked to perform a particular task when they hear a certain sound (eg placing a brick in a basket):

- The child must not be able to see the examiner making the test noise
- The sound stimulus may be warble tones or narrow-band noise at varying levels of intensity
- The child must understand and be familiar with the game first

Pure tone audiometry

A pure tone audiogram is a reliable method of testing children in each ear and can be consistently used in the normal child from 4–5 years onwards:

- Headphones are placed on the child and the test starts with an easily audible tone, gradually reducing until the tone can no longer be heard
- Each ear is tested in turn
- Pitfalls: uncooperative children, unintentional visual clues by the tester or the parent
- Anxious children may not respond to the sound even at high levels
- This should be cross-checked with formal or informal speech tests

Electrical response audiometry

These tests are based on the fact that a few milliseconds after sound reaches the ear, action potentials are propagated in the cochlea, along the auditory pathways to the brainstem and cortex. It is possible to detect these potentials and use them to find the child's threshold for hearing.

Brainstem auditory evoked responses (BSER)

The BSER is recorded from three surface electrodes (usually scalp, mastoid and forehead). It represents transmission of the auditory signal from the cochlea to the brainstem. The child can be awake (if very relaxed), asleep or under anaesthetic. The complex waveform is demonstrated by computer analysis of the EEG. It is particularly useful for children who are autistic, blind or severely learning disabled, and is widely used as a screening test in high-risk neonates.

13.5 Specific scenarios

Bottom shuffling

This is a common referral for paediatric assessment of development, as bottom shufflers are often not walking until 18–24 months. There is no evidence that bottom shuffling itself delays development in any other way; hence, it is important to establish the locomotor skills of a child who is over 18 months old and not walking but bottom shuffling. Bottom shuffling is usually an isolated event and such children will eventually walk, although a detailed history and examination is required, as children with hemiplegia often bottom shuffle.

Locomotor problems that could affect the lower limbs disproportionately (mimicking simple bottom shuffling)

- Spine pathology, eg spina bifida
- Neuromuscular problem: muscular dystrophy, spastic diplegia
- Bone/joint problem: developmental dysplasia of the hip, osteogenesis imperfecta
- Hypotonia

If you notice, or the parents report, bottom shuffling and the rest of the developmental examination is normal, be sure to mention that you would proceed to a full neurological examination and an examination of the hips and spine. You could also ask about a family history of bottom shuffling or late walking.

Handedness

Normally appears in third year.

Be aware of abnormal causes of hand preference before this age – these could include:

- Lower motor neurone lesions, eg brachial plexus injury (from birth trauma)
- Upper motor neurone lesions, eg hemiplegia
- Visual field problems
- Congenital abnormalities/musculoskeletal problems/trauma. Mention that you would want to take a detailed history including birth history and carry out a full neurological examination and assessment of vision as part of your summing up.

Child with severe global developmental delay

Children with severe delay have a lot of input to achieve developmental milestones, but the targets are the same as those for children who develop normally. They need to be able to move about, look around, explore the world with their senses, see, hear and communicate. They also need to develop independence with day-to-day activities, to play and to interact with people. Look for evidence of how they are being encouraged to do these things and it will give clues to their developmental stage and abilities:

- **Don't be thrown** – remember your structure. Comment on as much as you can observe and then ask if you may to talk to the parents about the rest
- **Gross motor** – try a 'head to toe' approach, thinking about head control, rolling, weight bearing, sitting independently, cruising and walking. Comment on any obvious aids to gross motor function – wheelchairs, specially adapted chair or walking aids
- **Fine motor/vision** – comment on obvious squints/glasses/eye contact. Do they reach/grasp/bring objects to the midline? Are they using both hands equally? Are they able to open their hands? Are they able to release objects they have grasped? Can they point?
- **Speech/hearing** – look for hearing aids. Describe the character/quality of their vocalisation. Ask whether they recognise familiar voices, respond to their own name, follow commands
- **Social** – think about toileting (are they in nappies?), eating and drinking (gastrostomy/nasogastric tube?). Do they help with dressing? Look for a social smile. Do they play with toys/explore their environment? How do they indicate what they want?

In your summing up, comment on whether the child is more delayed in one area than another and anything you have noticed that is being done to improve or aid their development. Try to assign a developmental age for each area you have examined. Make a comment on the limitations of your assessment. For example, if they are strapped into a wheelchair you may be unable to fully assess their locomotor function, or if they are not positioned upright with their head in the midline you may not be able to properly assess their fine motor function/vision.

Tired/uncooperative child

Be prepared for this. The examiner is looking for a sympathetic, opportunistic and imaginative approach! The parents may well be stressed. If the parents are not trying to distract the child out of their tantrum then it may be a sign that you will not be able to either. Don't try for too long. Comment on as much as you can observe. Be structured in your approach. Say 'can I ask the child's mother some questions?' and have a structured plan for which questions to ask.

Case – Uncooperative toddler

Maya is a 2-year-old girl who was upset throughout the examination today. I had difficulty engaging her in any activities to assess her fine motor and communication skills in particular.

In the area of social development she demonstrated some stranger anxiety when I entered the room. She appeared to be having a tantrum which is a normal developmental milestone for a 2-year-old. Her mother reports that she enjoys imitative play but does not understand sharing her toys. I note that she is wearing nappies and her mother reports that she is not yet dry by day. This gives her a social developmental age of 2 years.

She demonstrated running and climbing. She refused to try to kick a ball. Her mother reports that she has never pedalled a tricycle. She uses two feet per step when climbing stairs. Her gross motor skills have a developmental age of 2 years.

Maya's mother has no concerns about her vision. I was not able to assess her fine motor skills today but her mother reports that she can scribble, that she enjoys looking at picture books and can turn pages. She does not yet know any colours. This gives her a fine motor and visual developmental age of at least 2 years.

Maya did not answer me when I asked her questions about naming body parts. Her mother reports that she would normally be able to do this. She also knows her name and age but not her address. Her family understands her but her speech is often unintelligible to strangers. Her mother has no concerns about Maya's hearing. She has a developmental age of 2 years for speech and hearing.

Based on a developmental history from her mother, Maya has normal development across all areas. However, it was difficult to demonstrate this today due to her unwillingness to co-operate.

Autism spectrum disorder

Autism spectrum disorder (ASD) is becoming increasingly common with prevalence in the UK estimated at 1:100.

Boys:girls = 3:1.

ASD is a spectrum disorder; each child will have a different pattern of features individual to them. The common shared essential features are problems or abnormalities in the following three areas of development: social interaction, communication (verbal and non-verbal) and behaviour. These abnormalities are usually evident by the age of 3 years.

Social interaction

- Qualitative impairment of social interaction
- Difficulty in responding to social cues or in appreciating the feelings of others, and so failure to develop relationships with peers
- Can seem aloof and indifferent, although most show attachment to parents and carers
- Passive and in their own world until interrupted
- Slow to distinguish between parents and other adults

Communication (verbal and non-verbal)

- Delay or regression in speech development
- Limited attempts to communicate, ie by pointing or with facial expressions
- May avoid eye contact during communication
- Limited content of speech, pedantic and concrete interpretation of language
- Repetitive language. Echolalia is common
- Speech without intonation

Behaviour

- Reduced imagination, lack of pretend play
- Restrictive and repetitive range of behaviours and play
- Rigidity is imposed on day-to-day functioning and rituals are common
- Upset by minor changes in environment or routine
- Some imaginative play activities may be copied, eg from the television, and performed exactly the same repeatedly
- Specific repetitive behaviours which may be seen:
    - Twirling, rocking, head banging
    - Impulsivity, tantrums, over or under-activity

Suggested scheme for examination of the child with autism spectrum disorder

Children with ASD are commonly seen in the developmental section of the exam. You need to have a structured approach which assesses their development while looking for key features that point to the diagnosis.

Initial procedures

Introduce yourself to the parents and child.

Keep to the examiners' instructions. If asked to 'assess this child's development' then start with gross motor and fine motor, which should not be impaired in classic autism. This will give you time to watch their play and social interactions. If asked for just social or speech/hearing then start by saying as many things as you can observe that are relevant to these areas. If asked to assess whether a child has any autistic features then combine what you see with key questions for the parents.

Examination

Social

- Lack of stranger awareness
- Inability to direct child towards an activity
- Lack of eye contact
- You may not be able to get their attention at all
- Look for evidence of pretend play: how are they using the objects in the room and does it show imagination?
- Are they in nappies? (children with ASD often toilet-train later, though this is not a consistent finding).

Communication

- Are they using words with meaning? (If so, you can ask about name/age/body parts/colours, etc)
- Are they using facial expressions/gestures to communicate?
- How does their parent communicate with them?

Questions to ask

If you get an opportunity to ask questions then think about what you are trying to assess. You will not be expected to make a diagnosis of autism (a process that can sometimes take highly experienced people many hours of observation). However you should have an opinion as to whether further assessment for autism is indicated. You are trying to show a discrepancy in development between gross and fine motor skills and social/communication skills, and that the quality of social interaction and communication is not normal. You will need to demonstrate to the examiner that you have an understanding of what is normal development by the questions you ask. The following are questions you might ask:

1. Do you have any concerns about hearing or vision? (There may be a good reason why you can't get their attention)
2. Ask about toileting skills, dressing and eating
3. Ask about pretend play – given the choice, what or how would the child play?
4. Ask about friendships and sharing (in an older-looking preschool child)
5. Ask about language (vocabulary, putting words together, understanding commands/prepositions)
6. Ask how they communicate what they want; for example, children with ASD will often communicate they want a drink by taking their parents hand and using it to point to a cup.

These are crucial to making a sensible statement about your developmental examination. The following would be 'icing on the cake' if you have time to spare:

1. Ask about past medical history, family history
2. Ask about more specific 'autism' symptoms: rituals/repetitive behaviours/rigidity (remember that most normal toddlers feel safer with set routines and can have a tantrum when these are altered – is this child's behaviour more extreme than what you would normally expect?)

Finally

When summing up, order your findings in a structured way as described above. Make sure you compare delay in one area of development with the other areas and include some comment on the severity and the limitations of your assessment. Be sensitive, you will most likely be doing this in front of the child and parent.

Case – is an autism assessment indicated for this child?

Andrew is a 3-year-old boy with a family history of autism. On entering the room the most striking observation was that he did not appear to notice that we were there and did not react with any stranger anxiety or interest as is commonly seen in 3-year-old boys. He did not establish eye contact with anyone in the room, including his mother. He was constantly trying to turn the light switch on and off and showed no interest in the toys available. I was unable to engage him in any play activity, and he became distressed when his mother tried to move him away from the light switch.

He was vocalising but did not have any easily recognisable words and his mother reports that he indicates what he wants by taking her hand and putting it on the object he is interested in, such as the fridge or the television. He had very little facial expression.

The delay in social and communication skills was out of proportion to his motor development. I was unable to engage him in any formal assessment of these today; however I observed him walking and running and he was able to move a chair and climb onto it. These features would be in keeping with a diagnosis of autism. In addition to this, his mother describes some behavioural traits that are typical of children with autism spectrum disorder. In particular he has several rituals around dressing and washing. He becomes very anxious and distressed when there are small deviations from these rituals. Based on these findings I would agree that a referral for an autism assessment is indicated.

Down syndrome

A common misconception is that children with Down syndrome are grossly developmentally delayed. There is a huge spectrum of disabilities associated with Down syndrome and the disability tends to be a learning disability rather than a physical disability (unless the child has any of the conditions associated, such as congenital heart disease). In the first few years, development is often only slightly delayed and is a result of hypotonia. The learning problems become more apparent at 3–4 years. In view of this their educational placement should depend on their ability rather than their diagnosis – some children with Down syndrome enjoy and thrive on integration into mainstream schooling, although most will eventually require some form of more specialised provision to reach their full potential. When asked to carry out a developmental assessment on a child with Down syndrome the examination is exactly the same; they may fall within the normal range for motor development, so remember to report what you actually find!

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